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WE'RE WAY PAST PEAS: USES OF GENETIC INFORMATION TO UNDERSTAND HUMAN HEALTH AND GUIDE HEALTH CARE DECISION MAKING

Diana Nelson Louden, University of Washington Health Sciences Library Carolyn Martin, NN/LM Pacific Northwest Region

Introductions

Tell us about you:

□ Name

□ Job title

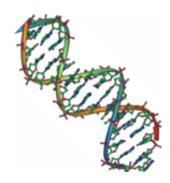
□ Place of work

City/State

Getting Into a Genetics Frame of Mind #1

How many people have had their DNA tested using a direct-to-consumer genetic test?

12 million +

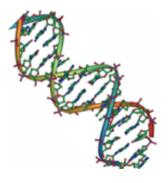


Getting Into a Genetics Frame of Mind #2

Which clinical specialists might use genetic information in patient care?

- a) Obstetricians
- b) Pediatricians
- c) Oncologists
- d) Cardiologists
- e) Pharmacists

All of them!

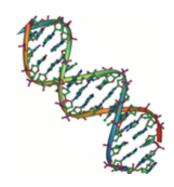


Getting Into a Genetics Frame of Mind #3

What is an example of a disease that could have a genetic component?

- a) Diabetes
- b) Breast cancer
- c) Osteoporosis
- d) Epilepsy
- e) Sickle-cell anemia

All of them COULD have a genetic component. Sickle-cell anemia is ALWAYS genetic.



Genomic Health Literacy

Genomics in the News

Scientists report CRISPR restores effectiveness of lung cancer treatment

A NEW GENETIC TEST COULD HELP DETERMINE CHILDREN'S SUCCESS

CRISPR gene editing can cause hundreds of unintended mutations

Baltimore Ravens to hand out

free DNA test kits

Can your DNA help you to make Technique That Combines New 3 Parents

Genetically Modified People Are Walking Among Us

Global study finds 44 genetic risk factors for major depression

Crime scene investigators couldn't tell identical twins' DNĂ apart. Until now

New Genetic Engineering Method Called Promising — And Perilous

Tackling the Opioid Crisis: Genetic testing to identify addiction risk

We will find you: DNA search used to nab Golden State Killer can home in on about 60% of white Americans Harnessing CRISPR for rapid detection of viral and

Genomic Literacy

- Genomic health literacy
- □ Genomic science literacy
- □ Role of media in genomic literacy

PMID: 23448722

PMID: <u>19341496</u>

Consumer Genomic Health Literacy

Lack biology basics

Lack mathematical concepts

Low health literacy



Definitions

- Genomic Health Literacy
 - The capacity to obtain, process, understand, and use genomic information for health related decision making.
- □ Genomic Science Literacy
 - The knowledge of basic genetics and genomics concepts and processes needed to build conceptual understanding, and the necessary mathematical knowledge to support this comprehension.

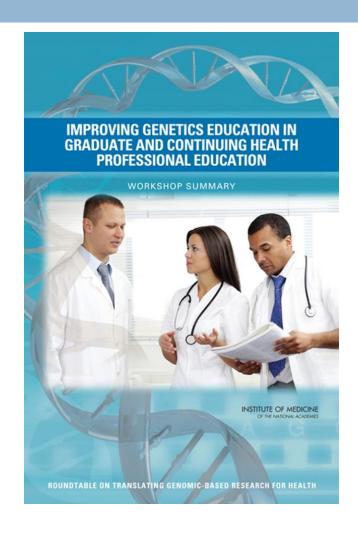
Leading causes of death

- 1. Heart disease: 635,260
- 2. Cancer: 598,038
- 3. Accidents (unintentional injuries): 161,374
- 4. Chronic lower respiratory diseases: 154,596
- 5. Stroke (cerebrovascular diseases): 142,142
- 6. Alzheimer's disease: 116,103
- 7. Diabetes: 80,058
- 8. Influenza and pneumonia: 51,537
- 9. Nephritis, nephrotic syndrome, and nephrosis: 50,046
- 10. Intentional self-harm (suicide): 44,965

Genomic Science Literacy

- □ K-12 education unable to keep up with scientific advancements
- Low emphasis on genomics
- Some teachers have misconceptions about genetics/genomics and little understanding
- Teachers need updated skills and have little access to genetic/genomic quality science curriculum
- Encourage partnerships with scientists
- Empower students entering the age of personal genomic medicine

Challenges for Clinicians – translating research to practice



"Despite the growing use of genomic applications in clinical practice, health professional knowledge about genomic information and confidence in using it have not kept pace....

Many health care providers do not have either the knowledge or the tools they need in order to apply genetic information in their day-to-day practices.

This lack of support is contributing to a substantial delay in the translation of genetic research findings, when appropriate, into improvement in patient outcomes within the health care system."

- Institute of Medicine 2015 report

Challenges for Clinicians: Translating Research to Practice

- Medical geneticists, genetic counselors, & advanced practice genetic nurses are the ones who are rigorously trained to deliver genetic health services.
- □ There's a shortage of genetic specialists.
- Primary care providers are on the "front line."
- There isn't sufficient evidence to rely on many genetic tests.
- New genetic variants are identified all the time;
 the significance of many of them is not known.
- Genomic information is not typically well integrated in electronic health records.



Challenges for Clinicians: Expanded Genetic Testing Capabilities

"I'm a community physician...I've got a medical license, I can order any blood test I want to, and I may or may not be well-trained in the precise implications of that test. Heck, I may not even be able to interpret the report because what used to be about a half page report has now grown to about a two or three page report and there's more BRCAs that you have to know the meaning of." — Breast Oncologist

Topics for Today

- Genomic health literacy
- Basic principles of genetics
- Uses of genetic information in health care
- NCBI's MedGen portal & other clinical genetics resources
- Practice answering questions using MedGen. Discussion.
 - Break 10:15-10:30
- Genetic consumer health resources
- Ethics and privacy
- Practice answering questions using Genetics Home Reference. Discussion.
 - Show what you know! Prizes! Evaluations!

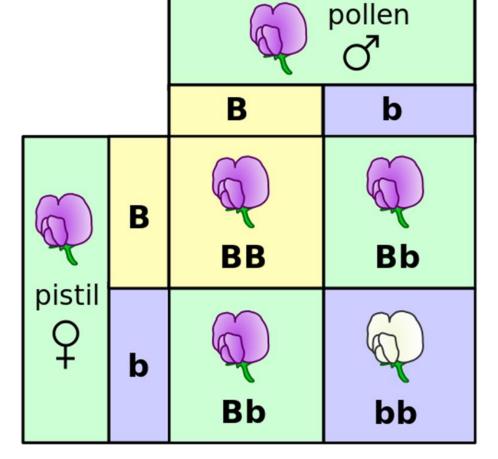
Presentation slides are available at: https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations

BUT FIRST, PEAS



Mendel Discovered Patterns of Inheritance by Studying Physical Traits

Before genes were discovered, Mendel realized that he could make mathematical predictions about the inheritance of physical traits – like flower color.



By Madprime (Own work) [CC0, GFDL (http://www.gnu.org/copyleft/fdl.html), CC-BY-SA-3.0 (http://creativecommons.org/licenses/by-sa/3.0/), via Wikimedia Commons

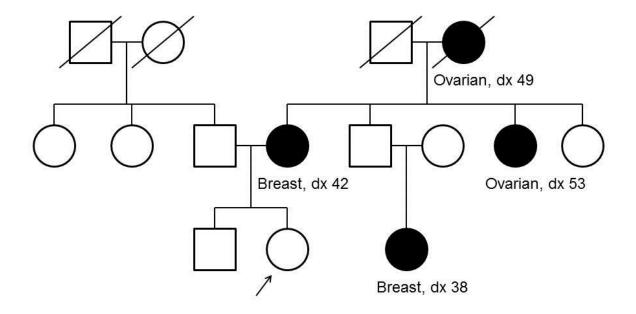
Mary-Claire King: Way Past Peas!



Mary-Claire King: Way Past Peas!

Demonstrated in 1990 that mutations in a single gene, which she named BRCA1, were responsible for breast and ovarian cancer in many families.

Classic BRCA1 Pedigree



Mary-Claire King: Using Genetic Information To Predict (and Prevent) Breast Cancer

- "Most of inherited breast and ovarian cancer can be prevented, if mutation carriers know who they are."
- □ But "about half of women who inherit mutations in BRCA1 or BRCA2 have no family history of breast or ovarian cancer and have no idea they are carrying cancer-causing mutations." [They've inherited the mutation from an "unaffected father."]
- Wants women to be offered genetic testing for BRCA1 and BRCA2 mutations at about age 30 as part of routine medical care.

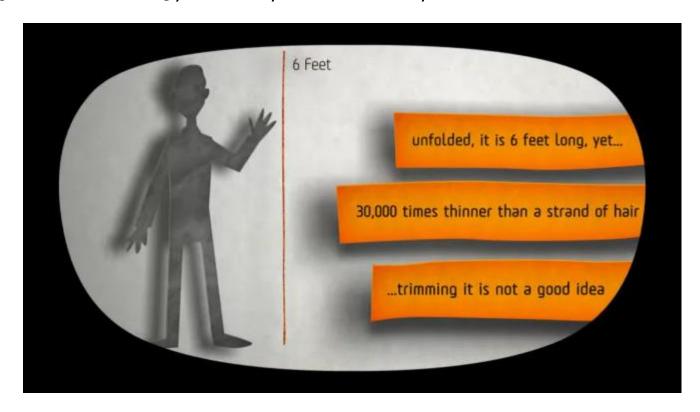
https://www.cnn.com/2019/02/19/health/brca-genetic-testing-recommendation-study/index.html

JUST ENOUGH GENETICS...

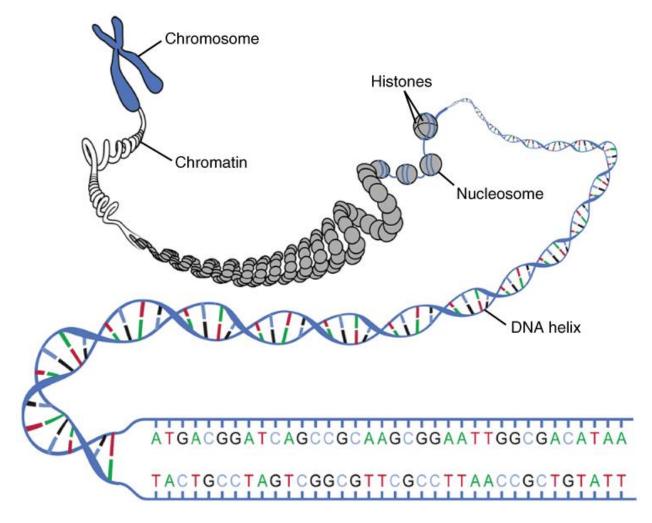
The Animated Genome

Unlocking Life's Code video

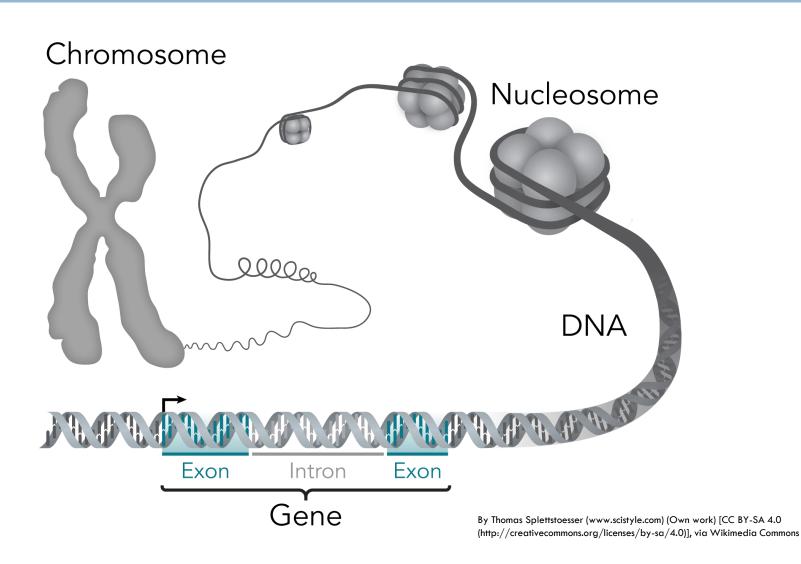
https://unlockinglifescode.org/media/animations/659#660



Chromosomes are made of DNA

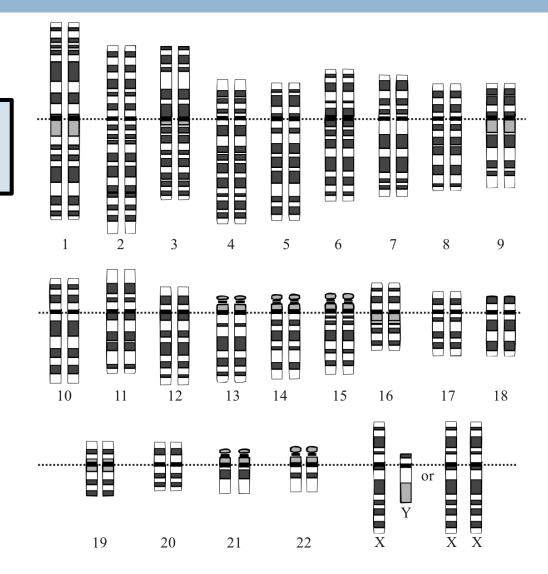


Genes are discrete segments of DNA found on chromosomes



Humans Receive 23 Chromosomes from Each Parent

Each of Your Cells Contains
These 46 Chromosomes*

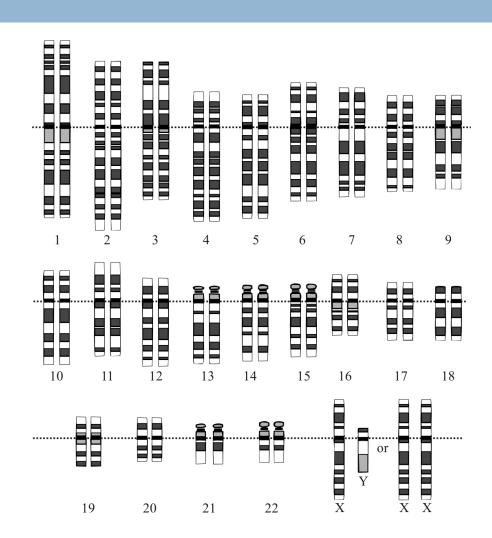


By Courtesy: National Human Genome Research Institute [Public domain], via Wikimedia Commons

^{*}Egg and sperm cells only contain one set of 23 chromosomes.

This is Your Genome

- The DNA that contains your
 20,000+ genes.
- The DNA that regulates the expression of your genes.
- The DNA of unknown significance.

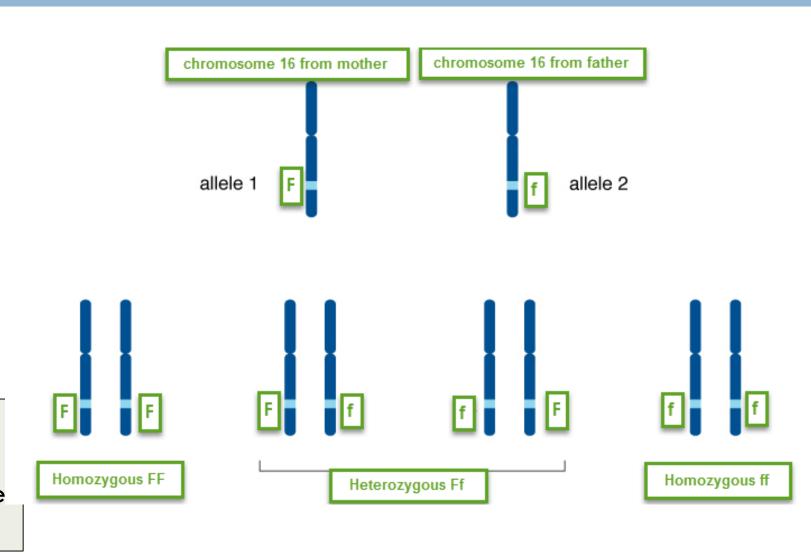


Genetic Alleles: Different Versions of the Same Gene



An allele is one of two or more versions/variants of a gene within a population.

"F" represents the freckle gene – MC1R. Freckles are a dominant trait. If you receive at least one copy of the F allele, you're likely to have freckles.



Many Traits Are Polygenic – the Product of Multiple Genes

Eye color is determined by variation at several different genes and the interactions between them.

Brown Eyes







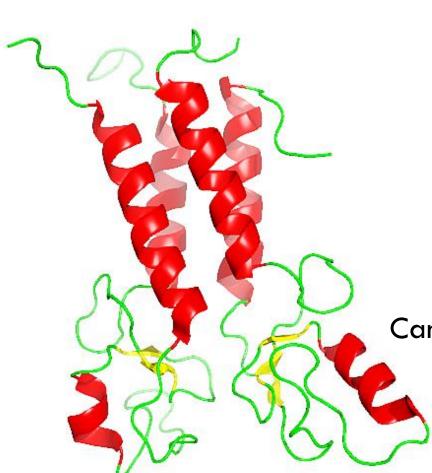
How does a gene affect a physical trait or process?

- Genes encode proteins.
- The DNA sequence of the gene dictates the amino acid sequence of the protein.
- A protein's sequence defines its 3-dimensional structure & how it can interact with other molecules
- □ Proteins do the work in
 your body.
 | Mage credit from Harvey Mudd College | web page: http://fourier.eng.htmc.edu/bioinformatics/

intro/node8.html

DNA strand 3' (template) TRANSCRIPTION **mRNA** Codon TRANSLATION Protein Trp Phe Gly Amino acid

Altered genes can lead to altered proteins. Altered proteins might not function properly.



Normal BRCA1 Protein

Job Description: Repair Damaged DNA

Job Location: Breast and Ovaries

Mutated BRCA1 Protein

Damaged DNA Not Repaired.

Mutations Accumulate.

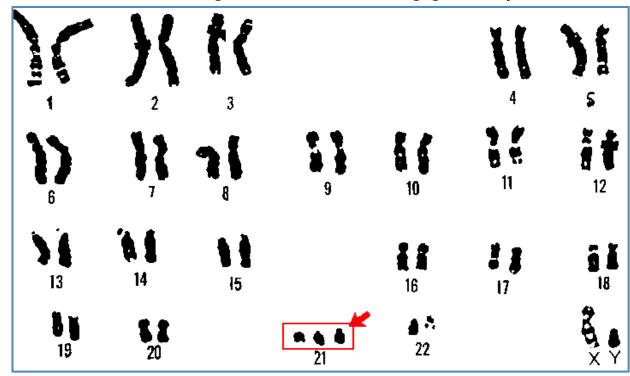
Can Lead to Uncontrolled Cell Growth and Tumor Formation

CATEGORIES OF DISEASES ATTRIBUTED TO GENES

- Chromosomal Diseases
- Monogenic Diseases/Mendelian Diseases
- Multifactorial Diseases

Chromosomal Diseases (Chromosomal Alterations)

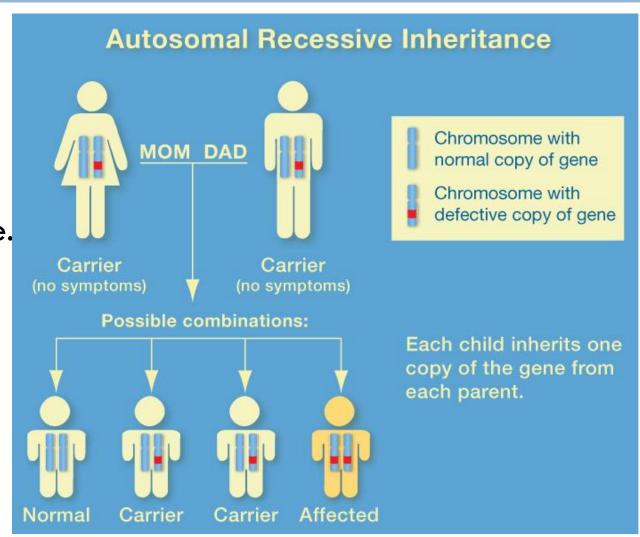
- An individual may have a missing chromosome, extra copies of a chromosome, or a portion of a chromosome may be deleted, duplicated, or translocated.
- Alteration may be inherited or de novo. Most originate in the egg or sperm.
- Example: Down's syndrome(extra copy of chromosome 21)



By U.S. Department of Energy Human Genome Program. [Public domain], via Wikimedia Commons

Monogenic Diseases/ Mendelian Diseases

- Single-gene diseases follow the patterns of inheritance that Mendel discovered in his studies of pea plants.
- These rare inherited diseases tend to be caused by mutations in a single gene.
- Examples: cystic fibrosis, sickle-cell anemia, muscular dystrophy, and Huntington's disease.



Multifactorial Diseases

- Complex diseases typically involve more than one gene.
- Also influenced by environmental factors, such as smoking, viral infections, and diet.
- Vast majority of human diseases fall into this category.
- Difficult to identify genes contributing to these diseases because a single physical condition can have many different causes.

Examples: cardiovascular disease, cancer, diabetes, and a number of birth defects

and psychiatric disorders.





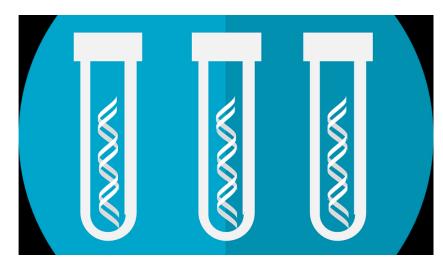
<u>Credit: Nature's Scitable website - http://www.nature.com/scitable/topic/genes-and-disease-17 NHGRI "Complex Disorders Background"</u>

https://www.genome.gov/10000865/

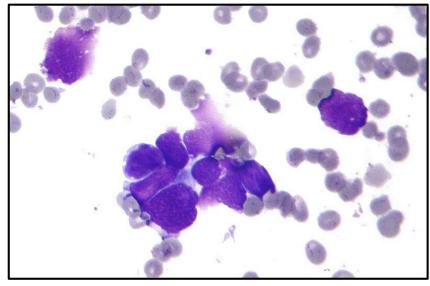
https://commons.wikimedia.org/wiki/File:Deep_fried_pineapple_on_a_stick_(2746967219).jpg

Genetic Risk Assessments Are NOT Psychic Predictions

Three Ways Genetics Is Intersecting Health Care

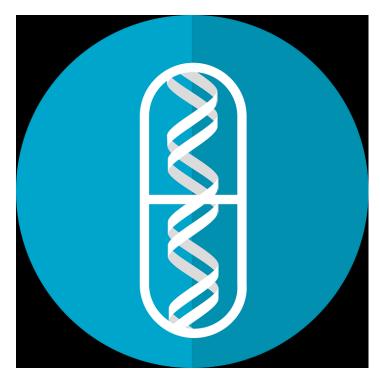


Genomic Testing



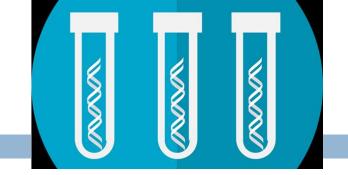
Credit: Wimedia Commons:

Tumor Testing



Pharmacogenomics

Clinical Genomic Testing (Germline Testing)



- Germline Testing: Mutations/variations found in all of your cells cells you've had since your beginning.
- Goals
 - Diagnosis can we attribute your observed condition to a genetic cause?
 - Risk assessment are you more likely than the average person to develop a particular disorder because of variation in particular genes?
- Types of Genetic Tests:
 - □ **Single gene tests** e.g., CFTR. [Mutations in both copies of CFTR cause cystic fibrosis.]
 - Gene panels multiple genes associated with a condition, e.g., colorectal cancer
 - Whole-exome sequencing & whole-genome sequencing looking for "something"

Clinical Genomic Testing in the Context of Patient Care

- Genomic testing is recommended after considering a patient's personal medical history and family medical history.
- Tests are selected based on determining what information is needed to guide patient care.
- Genetic tests are performed by academic and commercial labs.
- Clinician reviews test results and explains them to the patient.
- Next steps and management of patient care take genetic test results into consideration.

Vision for Genomic Medicine

"A vision for genomic medicine is that germline genome sequencing will be routinely conducted in health systems to provide healthcare and preventive services tailored to each individual [1].

For the most part, sequencing is not yet routinely used in clinical practice but is prioritized among people with certain diseases (e.g., ill newborns, and people with cancer or rare diseases) [2] or genetic predisposition to certain diseases (e.g., BReast CAncer susceptibility gene 1 [BRCA1] and BReast CAncer susceptibility gene 2 [BRCA2] testing for hereditary breast and ovarian cancer susceptibility) [3]."

Khoury MJ, Feero WG, Chambers DA, Brody LE, Aziz N, et al. (2018) A collaborative translational research framework for evaluating and implementing the appropriate use of human genome sequencing to improve health. PLOS Medicine 15(8): e1002631. https://doi.org/10.1371/journal.pmed.1002631

Classification Criteria

Tier 1

- . FDA label requires use of test to inform choice or dose of a drug
- FDA cleared or approved companion diagnostic device
- CMS covers testing
- Clinical practice guidelines based on systematic review supports testing

Tier 2

- FDA label mentions biomarkers
- FDA premarket approval (PMA)
- FDA 510(k) substantially equivalent decision
- CMS coverage with evidence development
- · Clinical practice guideline, not based on systematic review, supports use of test
- · Clinical practice guideline finds insufficient evidence but does not discourage use of test
- · Systematic review, without clinical practice guideline, supports use of test
- · Systematic review finds insufficient evidence but does not discourage use of test
- Clinical practice guideline recommends dosage adjustment, but does not address testing

Tier 3

- FDA label cautions against use
- · CMS decision against coverage
- · Clinical practice guideline recommends against use of test
- · Clinical practice guideline finds insufficient evidence and discourages use of test
- · Systematic review recommends against use
- · Systematic review finds insufficient evidence and discourages use
- Evidence available only from published studies without systematic reviews, clinical practice guidelines, FDA label or CMS labels coverage decision

CDC's Tier Database

CDC's Office of Public Health
Genomics (OPHG) ranks genomic
tests, and family health history
applications, by levels of evidence
phgkb.cdc.gov

The Answer to a Genetic Test Is Often Something Other Than Yes or No.

Labs typically report an individual's genetic test results using five categories

- 1. Disease-causing mutation found.
- 2. Mutation found is **likely** disease-causing.
- 3. Mutation found is **probably** benign and not disease causing.
- 4. Mutation is known to be benign and does not cause disease.
- 5. Mutation is a "Variant of **Unknown** Significance" (VUS).



Variants of Unknown Significance (VUSs)

Yes! A variant (SNP) is present in a gene. (We don't typically see that nucleotide in that location.)

No! We don't know what the clinical significance is. It could be benign, or it could be pathogenic.

A VUS in a lab report really is UNKNOWN.

After more evidence is collected, VUSs can often be categorized as benign or pathogenic.

Genetic Test Results Require Interpretation

- Clinicians don't order genetic tests unless the results are likely to improve patient management.
- Typically Medical Geneticists and Genetic Counselors are the clinicians who are most qualified to order and interpret genetic tests.

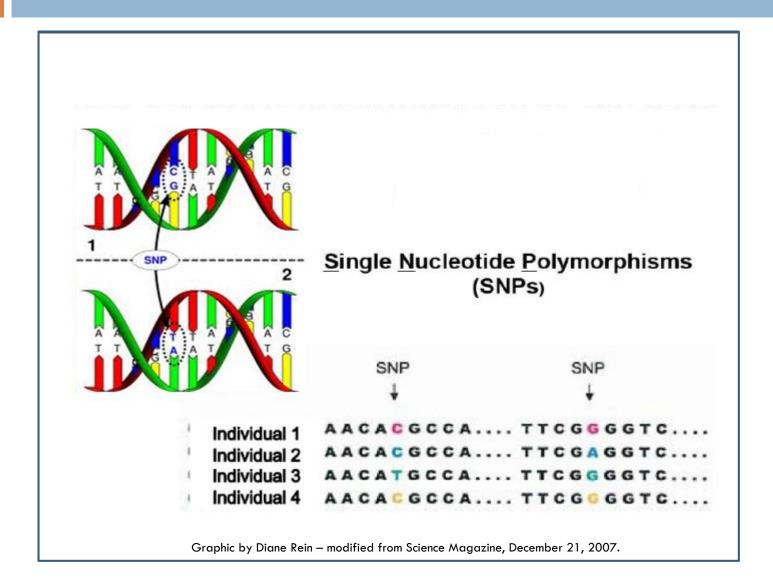


Polymorphism in Nature



By Debivort - Own work by Author, CC BY-SA 3.0, https://commons.wikimedia.org/w/index.php?curid=1626674

Clinical Genomic Tests Often Look for Single Nucleotide Polymorphisms (SNPs)





>2,600 SNPs Have Been Identified in the BRCA1 Gene >500 Variants are Considered "Causal"

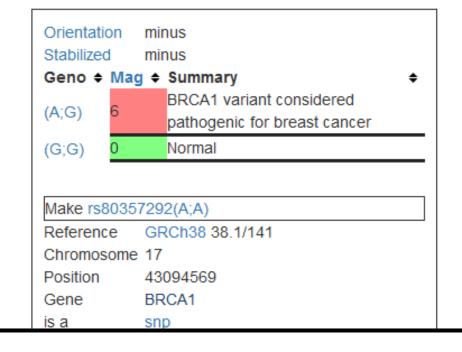
rs80357292

Here is one of those SNPs.

rs80357292, also known as W321X, c.962G>A and p.Trp321Ter, is a variant in the BRCA1 gene considered pathogenic for breast cancer in ClinVar.

Categories: Is a snp | In dbSNP | SNPs on chromosome 17 | Has genotype | On chip Ancestry v2 | On chip Ancestry v2c

In the BRCA1 gene — which is **193,689** nucleotides long — if this one particular nucleotide is an **A** instead of a **G**, a person is more likely to develop breast cancer.



https://www.snpedia.com/index.php/Rs80357292

BRCA1 SNPs -

"You have the breast cancer gene."

What should a person with a pathogenic BRCA1 variant expect?

- Increased risk of developing breast and/or ovarian cancer at an earlier age
- □ Lifetime risk of breast cancer of 80-90% (compared to 12% of women in the general population)
- Lifetime risk of ovarian cancer of 40-50% (compared to 1.3% of women in the general population)
- Increased risk of bilateral breast cancer

Clinicians Are Concerned with Clinically Actionable or Clinically Useful Genetic Test Results

- If you have a pathogenic BRCA1 variant, a clinician may be able to provide:
 - Closer surveillance (MRI in addition to mammogram)
 - Surgery (if warranted)
 - Chemoprevention
 - Genetic Counseling
- Sometimes the significance of a gene variant isn't known.
- Sometimes genetic testing is not likely to significantly improve patient outcomes and may lead to detrimental outcomes.

ACTIONABLE

23andMe Genetic Test for Breast Cancer Risk

- In 2018 FDA approved the marketing of the 23andMe "Personal Genome Service Genetic Health Risk Report for BRCA1/2 (Selected Variants)"
- Consumers receive a qualitative report on increased risk based on only <u>three</u> of the BRCA1 and BRCA2 mutations known to correspond to increased cancer risk



University of Michigan BRCA1 & BRCA2 Mutation Panel

GTR Home > Tests > BRCA1 and BRCA2 mutation panel

BRCA1 and **BRCA2** mutation panel

Clinical test of for Hereditary breast and ovarian cancer syndrome
Offered by Michigan Medical Genetics Laboratories

Test procedure 2

BRCA1 and BRCA2 Sanger sequencing of the entire coding region and deletion/duplication by MLPA

Variants Of Unknown Significance (VUS) Policy And Interpretation

What is the protocol for interpreting a variation as a VUS?
Follow ACMG guidelines, suggest family study

What software is used to interpret novel variations?
SIFT, PolyPhen2, RESCUE-ESE Web Server, Berkeley Drosophila Genome Project

What is the laboratory's policy on reporting novel variations?

Contact ordering physician by phone and by routine report; recommend clinical genetics follow up and/or genetic counseling and parental testing

GTR: GENETIC TESTING REGISTRY

Are family members with defined clinical status recruited to assess significance of VUS without charge?

No

Will the lab re-contact the ordering physician if variant interpretation changes?

Yes, by Phone or secure email

GTR Test ID 2: GTR000509980.1 Last updated: 2018-08-29

Test version history

- Looks for ALL variants in these two genes.
- Attempts to interpret any novel variants.
- If variant interpretation changes over time, lab will contact the ordering physician.

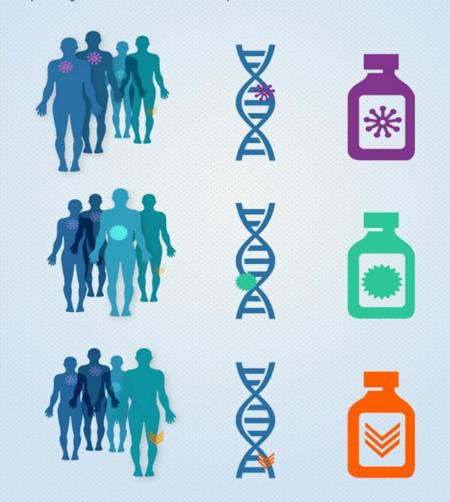
2. Tumor Testing: Identifying Genetic Mutations for Targeted Therapy

- Study a sample of tumor cells from a patient with cancer.
- Did an identifiable mutation lead to out-of-control cell growth?
- Do we have a drug that targets that specific out-of-control pathway?

PRECISION MEDICINE

NATIONAL CANCER INSTITUTE PRECISION MEDICINE IN CANCER TREATMENT

Discovering unique therapies that treat an individual's cancer based on the specific genetic abnormalities of that person's tumor.



Example of Tumor Testing Results



Genomics and Pathology Services

CORTEX Building, Suite 302,4320 Forest Park Ave. St. Louis, MO 63108 Phone: (314) 737-7337 | (866) 450-7697 | Fax: (314) 737-7336 Email: gps@wustl.edu | Website: gps.wustl.edu CLIA #26D0698285 | CAP #27556-03

PATIENT INFORMATION Name:

Date of Birth:

Gender:

Male

Disease: Adenocarcinoma of lung

Accession Number:

MR#:

Ordering Physician:

Two clinically significant genetic variants in EGFR (epidermal growth factor receptor)

identified in

patient's lung

cancer sample.

TEST PERFORMED



Solid Tumor Gene Set - Targeted next-generation sequencing was performed on this sample of Adenocarcinoma of lung. See Test Details for more information.

CLINICALLY RELEVANT RESULTS SUMMARY

Variants that are deemed clinically significant are listed here.

Variants Detected	In patient tumor type: FDA approved therapies, prognostic information, or other course of action	In another tumor type: FDA approved therapies, prognostic information, or other course of action
EGFR p.L747_T751del	✓	
EGFR p.T790M	✓	

Tumor Cells With Particular EGFR Mutations Can Be Treated With an EGFR Inhibiting Drug

Mutations are detected with an FDAapproved genetic test.

of these mutations, then recommended to treat with Tarceva. TARCEVA (erlotinib) tablets, for oral use Initial U.S. Approval: 2004

-----INDICATIONS AND USAGE-----

TARCEVA is a kinase inhibitor indicated for:

- The treatment of patients with metastatic non-small cell lung cancer (NSCLC) whose tumors have epidermal growth factor receptor (EGFR) exon 19 deletions or exon 21 (L858R) substitution mutations as detected by an FDA-approved test receiving first-line, maintenance, or second or greater line treatment after progression following at least one prior chemotherapy regimen. (1.1)
- First-line treatment of patients with locally advanced, unresectable or metastatic pancreatic cancer, in combination with gemcitabine. (1.2)

Limitations of Use:

- Safety and efficacy of TARCEVA have not been established in patients with NSCLC whose tumors have other EGFR mutations. (1.1)
- TARCEVA is not recommended for use in combination with platinumbased chemotherapy. (1.1)

3. Pharmacogenomics: Should We Prescribe You a Little, a Lot, or None At All?

Pharmacogenomics

Purpose:

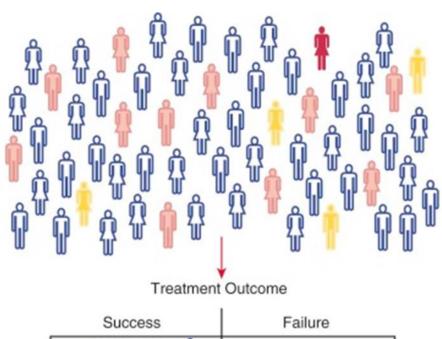
Study how genes affect an individual's responses to specific drugs.

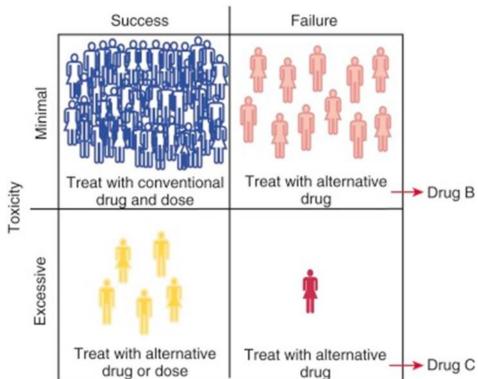
Goal:

Predict who will benefit from a medication, who will not respond, and who will experience adverse drug reactions.

Use that information to treat an individual with the appropriate drug and dose.

PRECISION MEDICINE





Adapted from Yaffe SJ, Aranda JV: Neonatal and pediatric pharmacology, 3rd ed., Philadelphia, 2004, Lippincott Williams & Wilkins

Plavix (Clopidogrel) and Pharmacogenomics

Plavix: anti-platelet drug; inhibits blood clots which could lead to heart attack and stroke

- Some people have genetic variants of an enzyme, CYP2C19, that cause them to metabolize Plavix more slowly.
- Slow metabolism of Plavix = increased risk of clotting/adverse events.
- Physicians will want to prescribe different anti-platelet drugs for these "poor metabolizers."



WARNING: DIMINISHED EFFECTIVENESS IN POOR METABOLIZERS

See full prescribing information for complete boxed warning.

- Effectiveness of Plavix depends on activation to an active metabolite by the cytochrome P450 (CYP) system, principally CYP2C19. (5.1)
- Poor metabolizers treated with Plavix at recommended doses exhibit higher cardiovascular event rates following acute coronary syndrome (ACS) or percutaneous coronary intervention (PCI) than patients with normal CYP2C19 function. (12.5)
- Tests are available to identify a patient's CYP2C19 genotype and can be used as an aid in determining therapeutic strategy. (12.5)
- Consider alternative treatment or treatment strategies in patients identified as CYP2C19 poor metabolizers. (2.3, 5.1)

FDA-Approved Drugs with Pharmacogenomic Information in Their Labels

Pharmacogenomics can play an important role in identifying responders and non-responders to medications, avoiding adverse events, and optimizing drug dose. Drug labeling may contain information on genomic biomarkers and can describe:

- Drug exposure and clinical response variability
- Risk for adverse events
- Genotype-specific dosing
- Mechanisms of drug action
- Polymorphic drug target and disposition genes
- Trial design features

As of April 2019, 261FDA-approved drugs have pharmacogenomics information in their labels.

Sometimes genetic testing is mandatory for prescribing a drug.

The table below lists therapeutic products from Drugs@FDA with pharmacogenomic information found in the drug labeling. The labeling for some, but not all, of the products includes specific actions to be taken based on the biomarker information. Pharmacogenomic information can appear in different sections of the labeling depending on the actions. For more information, please refer to the appropriate labeling guidance.

FDA's Table of Pharmacogenomic Biomarkers in Drug Labeling

Jean's Genetic Testing Timeline (get it, "Jean"?)

Age 1 day: Newborn testing for a few serious childhood diseases.

Age 30: Carrier testing for cystic fibrosis (with her partner) before trying to get pregnant. (Some members of extended family had CF.)

Age 35: Breast cancer risk prediction testing when her sister developed breast cancer at a young age.

Age 55: **Tumor testing** of breast cancer cells to determine the appropriate drug for treatment.

Age 70: **Pharmacogenomic testing** when Plavix (anti-platelet drug) was not effective.



TAKE A STRETCH BREAK!

Genetics and Medicine: Recommended Resources

Genetics and Medicine: Recommended Resources

Information resources on genetics and clinical practice, including genetic disorders, genetic testing, precision medicine, and pharmacogenomics

Genetics and Medicine Guide -

http://guides.lib.uw.edu/hsl/geneticmedicine

Genetic Medicine Resources: Starting Points for Clinicians

MedGen
MedGen







Genetic Testing Registry



PharmGKB



PubMed - Medical Genetics
Publimed

Clinical Genetics eBooks



Thompson and Thompson
Genetics in Medicine, 8th
ed. (2016) by Robert
Nussbaum; Roderick R.
McInnes; Huntington F.
Willard



Emery and Rimoin's
Principles and Practice of
Medical Genetics and
Genomics, 7th ed. (2019) by
Reed E. Pyeritz; Bruce R. Korf;
Wayne W. Grody

Key Genetic Medicine Resources

- MedGen
- o Summaries of medical genetics information compiled from GeneReviews, OMIM, ClinVar, Genetic Testing Registry, and PubMed.
- o Search for a gene, genetic disorder, or clinical feature.
- o Links to practice guidelines.
- · Gene Reviews
- o Point-of-care information for inherited conditions diagnosis, management, and genetic counseling information.
- o Peer-reviewed chapters typically focus on a single gene or phenotype/disorder.

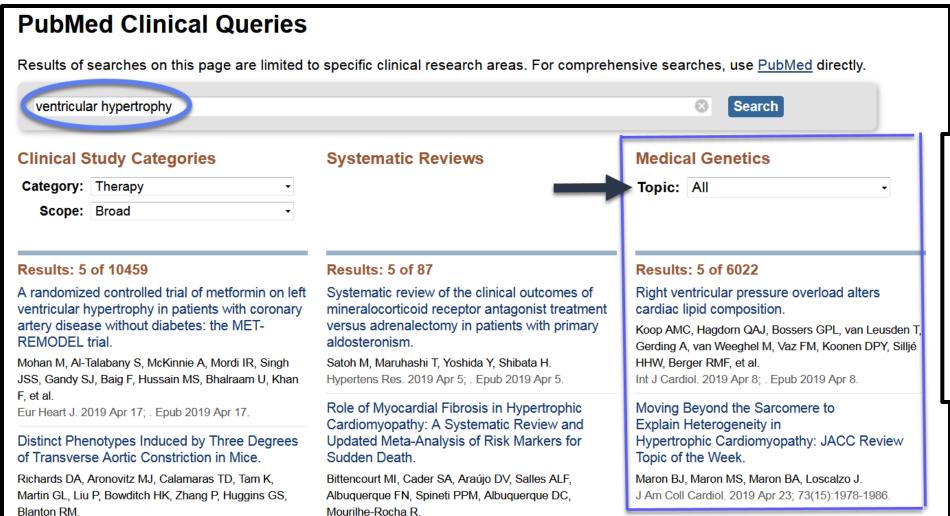
Medical Genetics Literature in PubMed

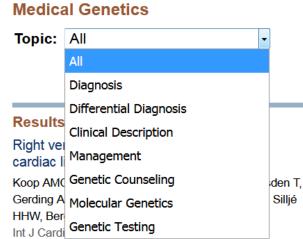
Two ways to efficiently identify PubMed references relating to medical genetics:

- Use the Medical Genetics clinical filter which can be found under "Clinical Queries"
- Locate a MeSH term that describes the topic you're interested in, and then add the "genetics" subheading. For example:
 - "Cardiomyopathy, Hypertrophic/genetics" [Mesh]

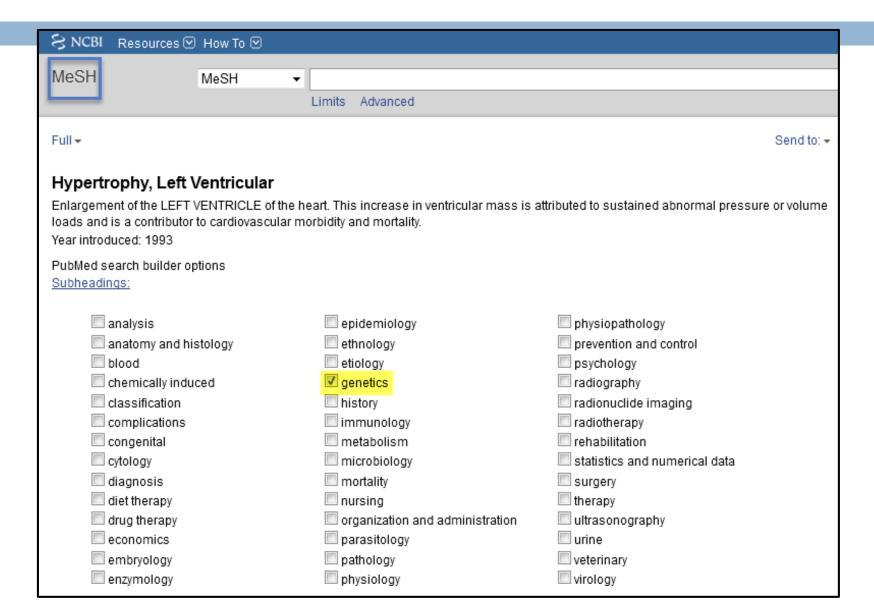
Genetic Tests

PubMed Medical Genetics Query





Genetics Subheading



Important Genetic Medicine Databases for Clinicians

	MedGen portal	Medical genetics information compiled from GeneReviews, OMIM, ClinVar, Genetic Testing Registry, practice guidelines, and PubMed. Links to consumer information.
	Gene Reviews	Point-of-care information for inherited conditions - diagnosis, management, and genetic counseling information. Peer-reviewed chapters. Search by gene or disorder.
	OMIM	Overviews of Mendelian disorders and genes associated with disease. Can search by symptom.
	GTR: Genetic Testing Registry	Tests for clinical use and genetic research. Individual genes and gene panels. Information submitted by test providers.
	ClinVar	Variants found in patient samples (primary data) along with assertions regarding the variants' clinical significance. Includes level of evidence available.
	PharmGKB	Information on the impact of human genetic variations on drug response. Includes drug dosing guidelines.

NCBI's MedGen Portal

http://www.ncbi.nlm.nih.gov/medgen





MedGen

Organizes information related to human medical genetics, such as attributes of conditions with a genetic contribution.

MedGen: NCBI Portal to Medical Genetics Content

- Single point of access to resources with information about human disorders and features/symptoms that have a genetic component.
- Designed for health care professionals & the medical genetics community.
- Integrates information from multiple sources: GeneReviews, OMIM, Genetic Testing Registry, ClinVar, Genetics Home Reference, professional guidelines, and PubMed.











GTR: GENETIC TESTING REGISTRY

MedGen: "One Stop Shop for Phenotypes with a Genetic Component"

Integrated content from multiple sources.

Professional guidelines – manually curated by NCBI staff

- □ Types of Records
 - Mendelian disorders
 - Pharmacogenetics of drug responses
 - Complex diseases with a genetic component
 - Clinical features

Follow Links from PubMed References to MedGen

J Am Coll Cardiol. 2019 Apr 9;73(13):1601-1611. doi: 10.1016/j.jacc.2018.12.085.

Cardiac Phenotypes, Genetics, and Risks in Familial Noncompaction Cardiomyopathy.

van Waning Jl¹, Caliskan K², Michels M², Schinkel AFL², Hirsch A³, Dalinghaus M⁴, Hoedemaekers YM⁵, Wessels MW¹, IJpma AS⁶, Hofstra RMW¹, van Slegtenhorst MA¹, Majoor-Krakauer D⁷.

Author information

Abstract

BACKGROUND: There is overlap in genetic causes and cardiac features in noncompaction cardiomyopathy (NCCM), hypertrophic cardiomyopathy (HCM), and dilated cardiomyopathy (DCM).

OBJECTIVES: The goal of this study was to predict phenotype and outcome in relatives according to the Send to: tases.

Cardiomyopathy (CMYO)

MedGen UID: 209232 · Concept ID: C0878544 · Disease or Syndrome

Synonyms: CMYO

Full Report -

SNOMED CT: Disorder of myocardium (57809008); Disorder of heart muscle (57809008);

Cardiomyopathy (85898001); Myocardiopathy (85898001); Myocardial disease (57809008)

Related genes: RBM20, NEXN, TMEM43, JPH2, MYOZ2, PRKAG2, ANKRD1, LDB3, ABCC9, BAG3, TCAP,

CSRP3, VCL, TTN, TPM1, TNNT2, TNNI3, TNNC1, TMPO, TGFB3, TAZ, SGCD, SDHA, SCN5A, RYR2, PSEN2, PSEN1, PLN, PKP2, MYL3, MYL2, MYH7, MYH6, MYBPC3, LMNA,

LAMA4, JUP, FKTN, EYA4, DSP, DSG2, DSC2, DES, CRYAB, ACTN2, ACTC1

HPO: HP:0001638 **Orphanet:** ORPHA167848

Definition

A disease of the heart muscle or myocardium proper. Cardiomyopathies may be classified as either primary or secondary, on the basis of etiology, or on the pathophysiology of the lesion: hypertrophic, dilated, or restrictive. [from NCI]

eening of relatives of 113 families from 143 index patients the cardiac phenotype. These cases were classified as dilation (DCM), and NCCM with LV hypertrophy (HCM).

tified 73 relatives with NCCM and 34 with DCM or HCM as higher in families with a mutation (p < 0.001). Fifty-four oserved in 37% of the relatives with a mutation. Index cases atives (p < 0.001). NCCM with DCM (53%) was associated ed risk for major adverse cardiac events, mutations in the tail in relatives (p < 0.001). Isolated NCCM (43%) was associated MYH7, asymptomatic NCCM (42%) (p = 0.018), and isolated M (4%) was associated with MYBPC3 and HCM without

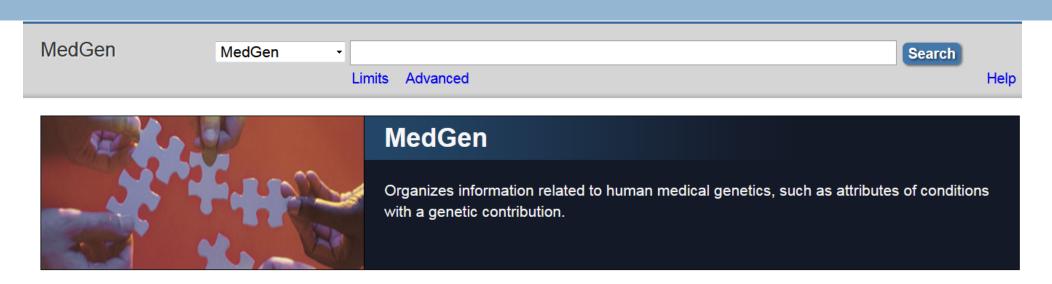
Go to: way be predicted according to the NCCM phenotype and the were related to outcome. In this way, clinical and genetic f outcome in relatives.

oundation. Published by Elsevier Inc. All rights reserved.



Best match search

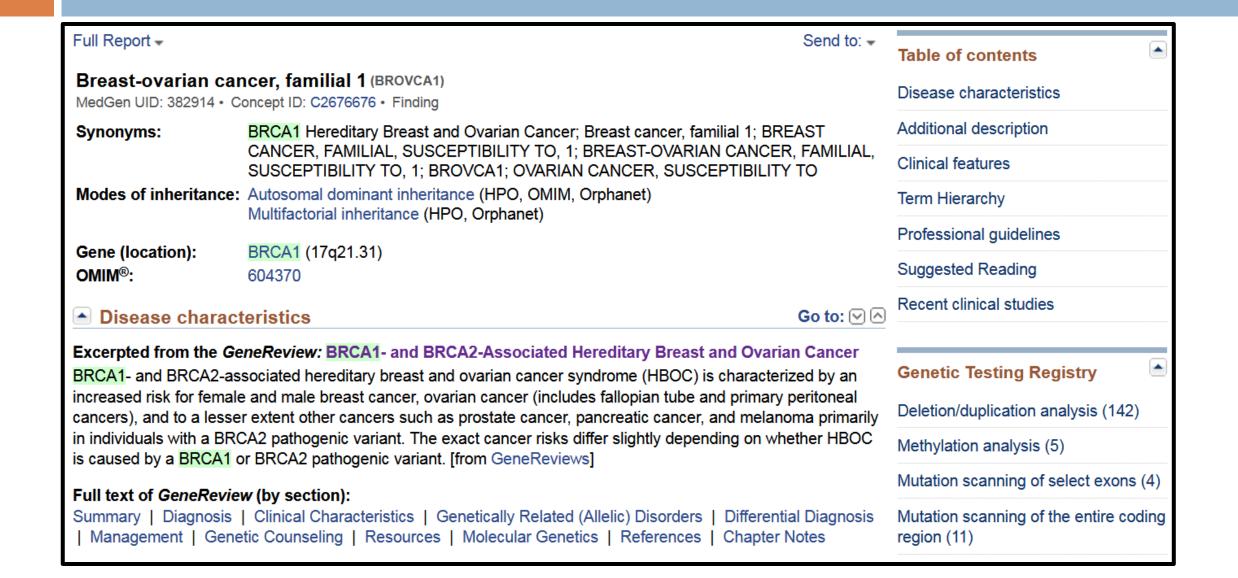
Exploration of MedGen Together



Example searches

Name	achondroplasia[title]	As you type your query, names of genetic disorders used in the NIH Genetic Testing Registry (GTR) will be provided. If you do not make a selection from the menu that appears under the search box as you type, your query is processed by looking for a match on a word or phrase. * is used as the wild card, and that wild card can be used only at the end of a word.
Related gene	LMNB1[gene]	If you enter a gene symbol followed by [gene], the diseases caused by or with some association to that gene will be retrieved.
Clinical feature	short stature[clinical features]	If you enter the name of the feature followed by [clinical feature] the diseases with that feature will be retrieved.

MedGen Summary. Search was BRCA1[gene]



Practice Question #1 Using MedGen

A patient is suspected to have the condition known as familial hypertrophic cardiomyopathy.

- Find an information-rich summary of familial hypertrophic cardiomyopathy (more general preferred). Make a note of the MedGen UID.
- What genes can be associated with familial hypertrophic cardiomyopathy?
- What year were the most recent professional guidelines written regarding the diagnosis and management of hypertrophic cardiomyopathy?
- According to Gene Reviews, mutations/pathogenic variants in what two genes cause most cases of familial hypertrophic cardiomyopathy?
- BONUS: According to the Genetic Testing Registry, is there a lab in the United States that offers a diagnostic panel (multiple genes) for hypertrophic cardiomyopathy?

Practice Question #2 Using MedGen

A physician suspects that her patient doesn't respond well to the drug clopidogrel (Plavix).

- 1) Find a MedGen record that addresses this phenomenon & note the UID.
- 2) What gene encodes the enzyme involved in metabolizing clopidogrel?
- When were the most recent professional guidelines published that discuss how to use clopidogrel pharmacogenetic information in practice?
- What percentage of Chinese people are thought to be poor metabolizers of clopidogrel? [Hint: Medical Genetics summaries link]
- 5) Can you find some information that may be helpful for the patient?
- 6) BONUS: What database has detailed information on the effects of gene variants on drug response? [Hint: It's linked from MedGen record.]

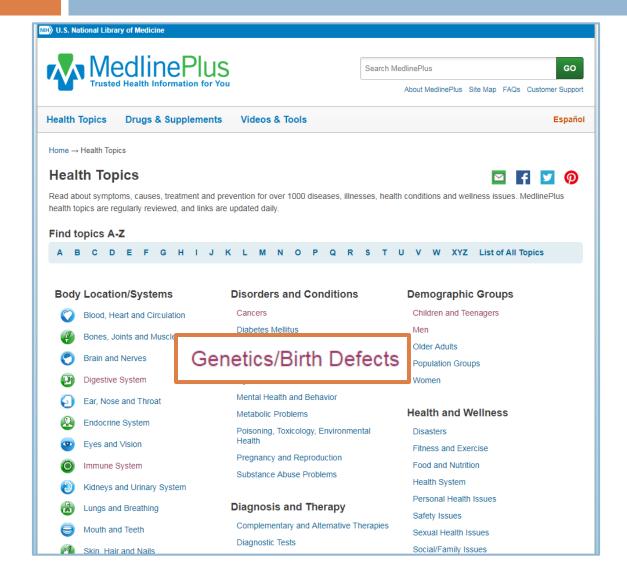
TAKE A BREAK!

PATIENT/CONSUMER GENOMIC HEALTH INFORMATION

The importance of being: genomically literate genomically informed

Patient/Consumer Resources

MedlinePlus



- Section: Genetics/BirthDefects
- □ Health Topic pages:
 - Genes and Gene Therapy
 - Genetic Brain Disorders
 - Genetic Counseling
 - Genetic Disorders
 - Genetic Testing
- text word search

MedlinePlus-Genetics/Birth Defects

Health Topics Drugs & Supplements Videos & Tools

Home → Health Topics → Genetics/Birth Defects

Genetics/Birth Defects

Abnormalities see Birth Defects

Achondroplasia see Dwarfism

Adrenoleukodystrophy see Leukodystrophies

Alpha-1 Antitrypsin Deficiency

Amniocentesis see Prenatal Testing

Anencephaly see Neural Tube Defects

Arnold-Chiari Malformation see Chiari Malformation

Ataxia see Friedreich's Ataxia

Ataxia Telangiectasia

Birth Defects

Blood Coagulation Disorders see Hemophilia

Brain Disorders, Inborn Genetic see Genetic Brain Disorders

Brain Malformations

Canavan Disease see Leukodystrophies

Cephalic Disorders see Brain Malformations

Cerebral Palsy

Charcot-Marie-Tooth Disease



Search MedlinePlus

About MedlinePlus Site Map FAQs Custom

Health Topics Drugs & Supplements Videos & Tools

Home → Health Topics → Heart Diseases

Heart Diseases

Also called: Cardiac diseases

On this page Learn More See, Play and Learn Basics Summary Living With Health Check Tools Start Here Related Issues Videos and Tutorials Diagnosis and Tests Specifics Prevention and Risk Factors Genetics Treatments and Therapies Resources For You Research Statistics and Research Reference Desk Children Clinical Trials Find an Expert Journal Articles Women Older Adults Patient Handouts

Summary

If you're like most people, you think that heart disease is a problem for others. But heart disease is the number one killer in the U.S. It is also a major cause of disability. There are many different forms of heart disease. The most common cause of heart disease is narrowing or blockage of the coronary arteries, the blood vessels that supply blood to the heart itself. This is called coronary artery disease and happens slowly over time. It's the major reason people have heart attacks.

Other kinds of heart problems may happen to the valves in the heart, or the heart may not pump well and cause heart failure. Some people are born with heart disease.

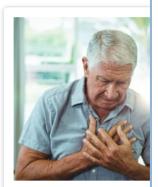
You can help reduce your risk of heart disease by taking steps to control factors that put you at greater risk:











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MEDICAL ENCYCLOPED

Aging changes in the heart and blood vessels

Aspirin and heart disease

- Silent Ischemia (Texas Heart Institute)
- Top 10 Myths about Cardiovascular Disease (American Heart Association)

Genetics

- · Genetic Testing for Inherited Heart Disease (American Heart Association)
- Genetics Home Reference: familial HDL deficiency (National Library of Medicine)
- Genetics Home Reference: Tangier disease NH) (National Library of Medicine)
- Genetics Home Reference: Williams syndrome NH (National Library of Medicine)

Health Check Tools

Heart-Health Risk Assessments from the American Heart Association (American Heart Association)

Videos and Tutorials

- Blood flow (Medical Encyclopedia)
 Also in Spanish
- Cardiovascular system (Medical Encyclopedia)
 Also in Spanish

Statistics and Research

- FastStats: Heart Disease (National Center for Health Statistics)
- · Heart Disease and Stroke Statistics (American Heart Association)
- . Interactive Atlas of Heart Disease and Stroke (Centers for Disease Control and Prevention)

Clinical Trials

- ClinicalTrials.gov: Cardiovascular Diseases NH (National Institutes of Health)
- ClinicalTrials.gov: Heart Diseases NH) (National Institutes of Health)

Journal Articles





Health Topics

Drugs & Supplements

Videos & Tools

Español

Home → Search Results

Related Topics

Seizures

Epilepsy

Ovarian Cancer

Breast Cancer

Genetic Counseling

Refine by Type

All Results (7,721)

- Health Topics (363)
- External Health Links (4,007)
- Drugs and Supplements (52)
- Medical Encyclopedia (448)
- MedlinePlus Magazine (143)
- Multiple Languages
- National Institutes of Health (2,514)

Genetic Disorders

Genes are the building blocks of heredity. parent to child. They hold DNA, the instru proteins. Proteins do most of the work in molecules from one place to another, buil toxins, and do many other maintenance jo

Sometimes there is a mutation, a change mutation changes the gene's instructions the protein does not work properly or is m called a genetic disorder.

(Read more

Results 1 - 10 of 5,178 for genetics

- Genetic Disorders (National Library
- ... This can course a medical condition c mutation from ... during your lifetime. The gene disorders, where a mutation affect https://medlineplus.gov/geneticdisorders
- Genetic Brain Disorders (National A genetic brain disorder is caused by a ... A mutation is a change in a gene. Ge

NIH) U.S. National Library of Medicine





Health Topics

Drugs & Supplements

Videos & Tools

Español

Search Help

Home → Search Results

Related Topics

Genetic Testing

Genes and Gene Therapy

Amyotrophic Lateral Sclerosis

Genetic Counseling

Refine by Type

All Results (1,701)

- Health Topics (2)
- External Health Links (847)
- Drugs and Supplements (10)
- Medical Encyclopedia (17)
- MedlinePlus Magazine (19)
- Multiple Languages
- National Institutes of Health (797)

Results 1 - 10 of 918 for genomics

- 1. **Genomics** and **Health Impact Update** (Centers for Disease Control and Prevention)
- ... Navigation in Pocs Home Facebook Twitter Email Syndicate **Genomics** and Precision Health Weekly Update Related Pages April 18, 2019 Spotlight **Genomics** and Population Health Action This week, we highlight ...

https://www.cdc.gov/genomics/update/current.htm - External Health Links

- 2. **Genomic Testing** (Centers for Disease Control and Prevention)
- ... TP Docs Home Facebook Twitter Email Syndicate Evaluating **Genomic** Tests Related Pages Fact Sheet: Identifying Opportunities to ... detail this database. Top of Page Evaluation of **Genomic** Applications in Practice and Prevention (EGAPPTM) In 2004, ... https://www.cdc.gov/genomics/gtesting/index.htm External Health Links
- 3. National Human Genome Research Institute (National Human Genome Research Institute)
- ... National Human Genome Research Institute conducts genetic and **genomic** research, funds genetic and **genomic** research and promotes that research to advance **genomics**

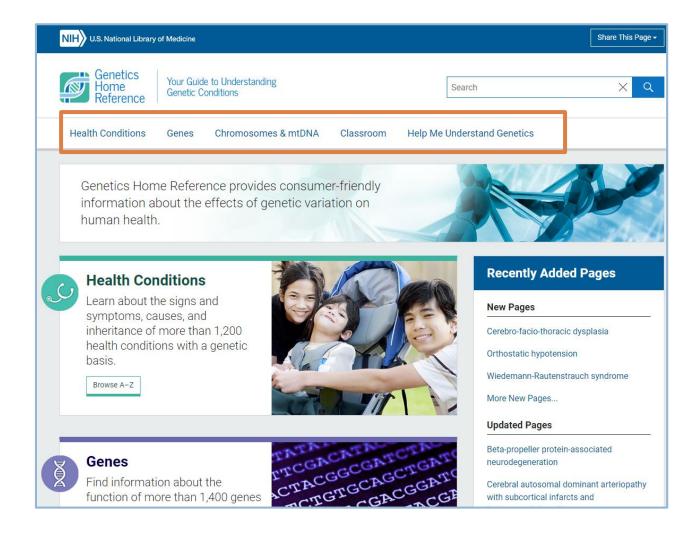
https://www.genome.gov - Other Resources

4. Frequently Asked Questions about Genetic and Genomic Science (National Human Genome Research Institute)

https://www.genome.gov/19016904 - External Health Links

Priof Guido to Conomics: DNA Gonos and Gonomos NH) (National Lluman

Genetics Home Reference (GHR)



- Health conditions
- Genes
- Chromosomes and DNA
- □ Classroom
- Help Me Understand Genetics





NIH U.S. National Library of Medicine

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Your Guide to Genetic Cond **Health Conditions**

Genes

Chromosomes & mtDNA

Classroom

Help Me Understand Genetics

Health Conditions

Health Conditions

Genes



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Health C

Explore the sig of various heal







B variant GM2 ganglio B. burgdorferi infectior

Baller-Gerold syndrom

Ballinger-Wallace synd

Baltic myoclonic epiler

Baltic myoclonus, see Baltic myoclonus epile

BAM syndrome, see Bo

bamboo hair syndrome

BAMS, see Bosma arhi

BANF, see Neurofibron

bangungut, see Brugad

Bannayan-Riley-Ruvalo

Bannayan-Ruvalcaba-F

Bannayan-Zonana syn

Breast cancer

▶ Description

▶ Frequency

▶ Causes

▶ Inheritance Pattern

▶ Diagnosis & Management Linl

▶ Other Names for This Condition

▶ Additional Information & Resc

▶ Sources for This Page

▶ Frequency

▶ Description

▶ Causes

▼ Inheritance Pattern

Most cases of breast cand associated with somatic they do not cluster in fami

In hereditary breast cance example, mutations in the pattern , which means or chance of developing cand mutated gene can be inhe

In the other syndromes dis autosomal dominant patte likelihood of developing ca genes will ultimately devel

In many cases of breast c mechanism of inheritance ▼ Additional Information & Resources

▶ Health Information from MedlinePlus (3 links)

▶ Genetic and Rare Diseases Information Center (2 links)

► Additional NIH Resources (4 links)

▶ Educational Resources (9 links)

▶ Patient Support and Advocacy Resources (10 links)

▶ Clinical Information from GeneReviews (5 links)

▶ Scientific Articles on PubMed (1 link)

▶ Catalog of Genes and Diseases from OMIM (3 links)

▶ Medical Genetics Database from MedGen (2 links)

▶ Diagnosis & Management Links

▶ Other Names for This Condition



Health Conditions

Genes

Chromosomes & mtDNA

Classroom

Help Me Understand Genetics



Learn More about Genes

What is DNA?

Explore the normal functions of human genes and the health

implications of genetic changes.

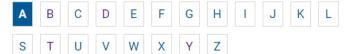
Health Conditions

Genes

Chromosomes & mtDNA

Classroom

Help Me Understand Genetics



AAAS: aladin WD repeat nucleoporin

AASS: aminoadipate-semialdehyde synthase

ABAT: 4-aminobutyrate aminotransferase

ABCA1: ATP binding cassette subfamily A member 1

ABCA3: ATP binding cassette subfamily A member 3

ABCA4: ATP binding cassette subfamily A member 4

ABCA12: ATP binding cassette subfamily A member 12

ABCB4: ATP binding cassette subfamily B member 4

ABCB7: ATP binding cassette subfamily B member 7

ABCB11: ATP binding cassette subfamily B member 11

ABCC2: ATP binding cassette subfamily C member 2

ABCC6: ATP binding cassette subfamily C member 6

ABCC8: ATP binding cassette subfamily C member 8

ABCC9: ATP binding cassette subfamily C member 9

ABCD1: ATP binding cassette subfamily D member 1

ABCD4: ATP binding cassette subfamily D member 4

ABCG2: ATP binding cassette subfamily G member 2 (Junior blood group)

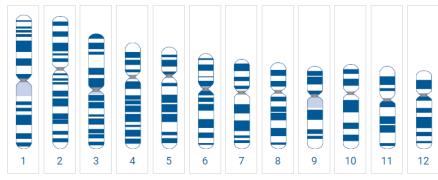
ABCG5: ATP binding cassette subfamily G member 5

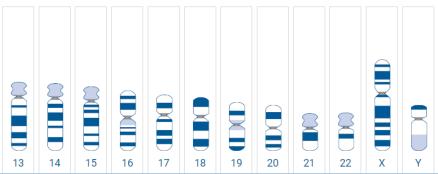
ABCG8: ATP binding cassette subfamily G member 8



Read about each of the human chromosomes and mitochondrial DNA (mtDNA) and the health implications of genetic changes.

Information about specific chromosomes





Learn More about Chromosomes & mtDNA

What is a chromosome?

How many chromosomes do people have?

Can changes in the number of chromosomes affect health and development?

Can changes in mitochondrial DNA affect health and development?



Help Me Understand Genetics

An introduction to fundamental topics related to human general genetics concepts.

- ▶ Cells and DN
- ▶ Mutations an
- ▶ How Genes V
- ▶ Gene Groups
- ▶ Inheriting Ge
- ▶ Genetics and
- ▶ Genetic Cons
- ▶ Genetic Testi
- ▶ Direct-to-Cor
- ▶ Newborn Scr
- ▶ Gene Therapy
- ▶ The Human Genome Project
- **▶** Genomic Research
- **▶** Precision Medicine

▼ Inheriting Genetic Conditions

Chromosomes & mtDNA

What does it mean if a disorder seems to Why is it important to know my family m What are the different ways in which a ge If a genetic disorder runs in my family, w What are reduced penetrance and variab What do geneticists mean by anticipation What are genomic imprinting and unipare Are chromosomal disorders inherited? Why are some genetic conditions more c

What is heritability?

- **Health Conditions** Genes
- Chromosomes & mtDNA

Classroom

Help Me Understand Genetics

If a genetic disorder runs in my family, what are the chances that my children will have the condition?

When a genetic disorder is diagnosed in a family, family members often want to know the likelihood that they or their children will develop the condition. This can be difficult to predict in some cases because many factors influence a person's chances of developing a genetic condition. One important factor is how the condition is inherited. For example:

- Autosomal dominant inheritance: A person affected by an autosomal dominant disorder has a 50 percent chance of passing the mutated gene to each child. The chance that a child will not inherit the mutated gene is also 50 percent. However, in some cases an autosomal dominant disorder results from a new (de novo) mutation at that occurs during the formation of egg or sperm cells or early in embryonic development. In these cases, the child's parents are unaffected, but the child may pass on the condition to his or her own children.
- Autosomal recessive inheritance: Two unaffected people who each carry one copy of the mutated gene for an autosomal recessive disorder (carriers) have a 25 percent chance with each pregnancy of having a child affected by the disorder. The chance with each pregnancy of having an unaffected child who is a carrier of the disorder is 50 percent, and the chance that a child will not have the disorder and will not be a carrier is 25 percent.
- X-linked dominant inheritance: The chance of passing on an X-linked dominant condition differs between men and women because men have one X chromosome and one Y chromosome, while women have two X chromosomes. A man passes on his Y chromosome to all of his sons and his X chromosome to all of his daughters. Therefore, the sons of a man with an X-linked dominant disorder will not be affected, but all of his daughters will inherit the condition. A woman passes on one or the other of her X chromosomes to each child. Therefore, a woman with an X-linked dominant disorder has a 50 percent chance of having an affected daughter or son with each pregnancy.
- X-linked recessive inheritance: Because of the difference in sex chromosomes, the probability of naccing on an Y-linked recessive disorder 🔯 also differs between men and women. The sons of a

For more information about passing on a genetic disorder in a family:

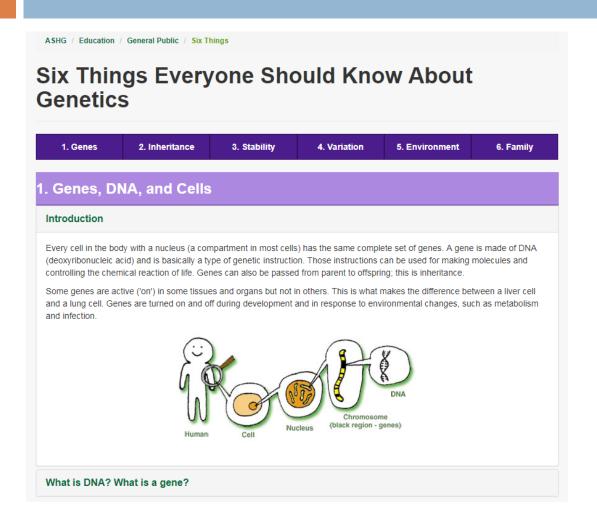
The National Library of Medicine MedlinePlus website offers information about the chance of developing a genetic disorder on the basis of its inheritance pattern:

- Autosomal dominant
- Autosomal recessive
- X-linked dominant
- X-linked recessive

The Centre for Genetics Education provides an explanation of mitochondrial inheritance .

The Muscular Dystrophy Association explains patterns and probabilities (PDF) & of inheritance.

Learning about genetics

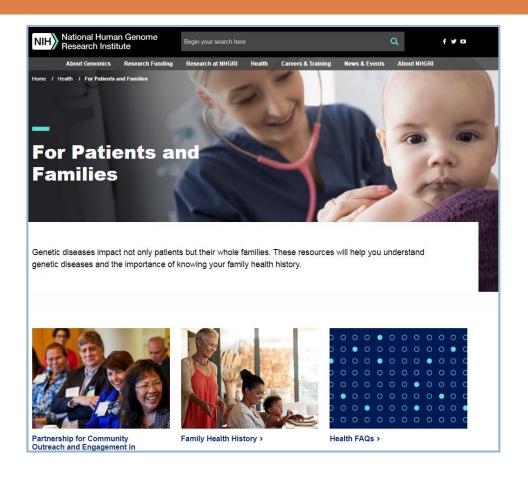


- 1) Genes
- 2) Inheritance
- 3) Stability
- 4) Variation
- 5) Environment
- 6) Family

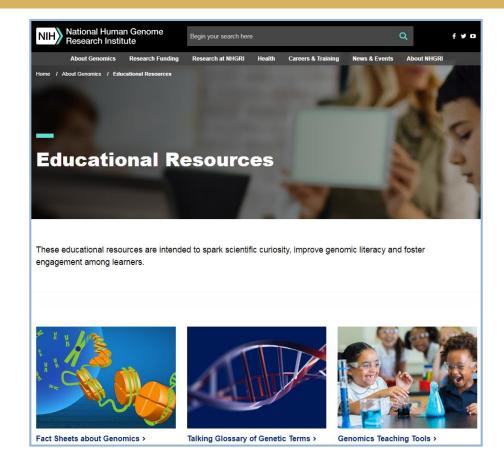
Six Things Everyone Should Know About Genetics

National Human Genome Research Institute (NHGRI)

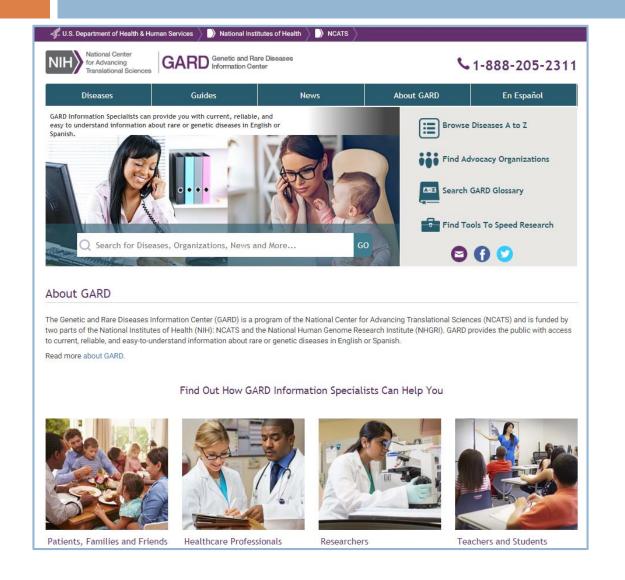
NHGRI Information for Patients & Public



NHGRI Education Resources



Genetic and Rare Diseases Information Center (GARD)



- □ How to Find a Disease Specialist
- Tips for the Undiagnosed
- Support for Patients and Families
- Tips for Finding Financial Aid
- Help with Travel Costs
- How to Get Involved in Research
- FAQs About ChromosomeDisorders
- Medical and Science Glossaries

NIH Institutes, Centers, and Offices

Quick Links

NICHD

NIGMS

NINDS

NIAAA

NIDCR

NIEHS

NIMHD

NCATS NCCIH

NTAMS

NIDCD

NIDA

NIMH

NINR

INSTITUTES, CENTERS, AND OFFICES

Institutes at NIH

List of NIH Institutes, Centers, and Offices

Directors of NIH Institutes and Centers

NIH Institute and Center Contact Information

NIH Office of the Director

List of NIH Institutes, Centers, and Offices



NIH Offices

NIH Office of the Director (OD)

The Office of the Director is the central office at NIH for its 27 Institutes and Centers. The OD is responsible for setting policy for NIH and for planning, managing, and coordinating the programs and activities of all the NIH components. OD program offices include the Office of AIDS Research and the Office of Research on Women's Health, among others.

NIH Institutes

National Cancer Institute (NCI) — Est. 1937

NCI leads a national effort to eliminate the suffering and death due to cancer. Through basic and clinical biomedical research and training, NCI conducts and supports research that will lead to a future in which we can prevent cancer before it starts, identify cancers that do develop at the earliest stage, eliminate cancers through innovative treatment interventions, and biologically control those cancers that we cannot eliminate so they become manageable, chronic diseases.

National Eye Institute (NEI) — Est. 1968

The National Eye Institute's mission is to conduct and support research, training, health information dissemination, and other programs with respect to blinding eye diseases, visual disorders, mechanisms of visual function, preservation of sight, and the special health problems and requirements of the blind.

National Heart Tung and Blood Institute (NHLBI) - Est 1948

- National Cancer Institute
- National Heart, Lung, and Blood Institute
- Eunice Kennedy Shriver National Institute of Child Health and Human Development
- National Institute of Neurological
 Disorders and Stroke
- □ etc...

NIH Institutes, Centers and Offices

National Organization for Rare Disorders (NORD)



- For Patients and Families
 - rare disease information
 - information to give to physicians
 - health insurance information
 - financial assistance
 - support and advocacy

NORD

Genetic Counseling Cultural Competence Toolkit

A GENETIC COUNSELING CULTURAL COMPETENCE TOOLKIT



Home About Us Background Health Disparities Cross Cultural Communication Cases Teaching Tools Clinical Tools CEUs Site Map Contact Us

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Search

Welcome to the Genetic Counseling Cultural and Linguistic Competence Toolkit

The Genetic Counseling Cultural and Linguistic Competence Toolkit (GCCCT) is the final product of the 2009 Jane Engelberg Memorial Fellowship (JEMF) award of the National Society of Genetic Counselors. This award facilitated professional development opportunities for one member of the genetic counseling profession, Nancy Steinberg Warren, MS, CGC in diversity, cultural competence, and pedagogy. The resulting project outcome is this website, which is designed as a flexible, online resource portal to benefit the entire genetic counseling profession.



Feedback from Our Users & What's New?

Learn about our CONSULTING and TRAINING services!

Continuing Education Units

The full 9-case registration fee is now only \$200! (\$100 off the usual registration fee of \$300.)

Genetic Counselors and Nurses: The National Society of Genetic Counselors has authorized approval of up to 1.44 CEUs or 14.43 contact hours (Category 1) for the Cases section of the Toolkit (9 cases).

Medical Interpreters: The International Medical Interpreters Association has authorized approval of 1.44 IMIA Continuing Education Units for the Cases section of the Toolkit.

Translators: The American Translators Association has authorized approval of 10 ATA Continuing Education Points for the Cases section of the Toolkit.

- □ For Genetic Counselors
- Offers CE
 - National Society of Genetic Counselors
 - American Board of Genetic Counseling
 - International Medical Interpreters
 Association
 - American Translators Association

Genetic Counseling Cultural Competence Toolkit

Addressing Specific Diagnosis

Sickle Cell Disease



Health Topics Drugs & Supplements

Home → Health Topics → Sickle Cell Disease

Sickle Cell Disease

Also called: Hemoglobin SS disease, Sickle cell

Learn More

Living With

Genetics

Resources

Find an Exper

Related Issues

On this page

Basics

Summary

- Start Here
- Diagnosis and Tests
- Treatments and Therapies

Research

- Statistics and Research
- Clinical Trials
- Journal Articles

Summary

What is sickle cell disease?

Sickle cell disease (SCD) is a group of inherited red problem with your hemoglobin. Hemoglobin is a pro throughout the body. With SCD, the hemoglobin for changes the shape of the red blood cells. The cells them into a crescent, or sickle, shape.

The sickle-shaped cells are not flexible and cannot they move through your blood vessels. The sickle c normal 90 to 120 days. Your body may have trouble you lost. Because of this, you may not have enough and it can make you feel tired.

NIH: National Heart, Lung, and Blood Institute

Start Here

- · Facts about Sickle Cell Disease (Centers for Disease Control and Prever Also in Spanish
- . Sickle Cell Disease (American Academy of Family Physicians) Also in Spanish
- Sickle Cell Disease (Centers for Disease Control and Prevention)
- · Sickle Cell Disease (SCD) (National Marrow Donor Program)
- What is Sickle Cell Disease? NH) (National Heart, Lung, and Blood Institute Also in Spanish
- When Blood Cells Bend: Understanding Sickle Cell Disease NH) (I)

Diagnosis and Tests

- Blood Smear NIH (National Library of Medicine) Also in Spanish
- Complete Blood Count (CBC) NIH (National Library of Medicine) Also in Spanish
- Hemoglobinopathy Evaluation (American Association for Clinical Chemist
- Sickle Cell Tests (American Association for Clinical Chemistry)

Treatments and Therapies

- Blood and Bone Marrow Transplant? NH) (National Heart, Lung, and B)
- Bone Marrow Transplantation: MedlinePlus Health Topic NH (National) Also in Spanish
- Complications and Treatments (Centers for Disease Control and Preventi Also in Spanish

Living With

- . Living Well with Sickle Cell Disease (Centers for Disease Control and Pr Also in Spanish
- · Sickle Cell Crisis (For Teens) (Nemours Foundation) Also in Spanish

Related Issues

- . Five Tips to Help Prevent Infections (Centers for Disease Control and Pr Also in Spanish
- For People of African, Mediterranean, or Southeast Asian Heritage:

Genetics

- Genetics Home Reference: sickle cell disease NH) (National Library of Medicine)
- Learning about Sickle Cell Disease NH) (National Human Genome Research Institute)
- Prenatal Genetic Counseling (For Parents) (Nemours Foundation) Also in Spanish
- Sickle Cell Trait (Centers for Disease Control and Prevention) Also in Spanish

Health Check Tools

· Sickle Cell Disease Quiz (Centers for Disease Control and Prevention) Also in Spanish

Videos and Tutorials

· What Is Sickle Cell Disease? (Dolan DNA Learning Center)

Statistics and Research

. Data and Statistics (Centers for Disease Control and Prevention)

Clinical Trials

ClinicalTrials.gov: Anemia, Sickle Cell NIH) (National Institutes of Health)

Journal Articles

References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- · Article: Knowledge Deficit of Sickle Cell Trait Status: Can Nurses Help?
- Article: Inhaled steroids associated with decreased macrophage markers in nonasthmatic individuals with...
- Article: Association of Matched Sibling Donor Hematopoietic Stem Cell Transplantation With Transcranial.
- Sickle Cell Disease -- see more articles

Find an Expert

- · Find a Genetic Counselor (National Society of Genetic Counselors)
- National Heart, Lung, and Blood Institute NH)
- · Sickle Cell Disease Association of America

Children



Genes

Chromosomes & mtDNA

Classroom

Help Me Understand Genetics

Sickle cell disease



Health Conditions

Genes Chromosomes & mtDNA

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► Frequency

▼ Causes

Mutations in the HBB gene cause sickle cell disease.

Hemoglobin consists of four protein subunits, typically, two subunits called alpha-globin and two subunits called beta-globin. The *HBB* gene provides instructions for making beta-globin. Various versions of beta-globin result from different mutations in the *HBB* gene. One particular *HBB* gene mutation produces an abnormal version of beta-globin known as hemoglobin S (HbS). Other mutations in the *HBB* gene lead to additional abnormal versions of beta-globin such as hemoglobin C (HbC) and hemoglobin E (HbE). *HBB* gene mutations can also result in an unusually low level of beta-globin; this abnormality is called beta thalassemia.

In people with sickle cell disease, at least one of the beta-globin subunits in hemoglobin is replaced with hemoglobin S. In sickle cell anemia, which is a common form of sickle cell disease, hemoglobin S replaces both beta-globin subunits in hemoglobin. In other types of sickle cell disease, just one beta-globin subunit in hemoglobin is replaced with hemoglobin S. The other beta-globin subunit is replaced with a different abnormal variant, such as hemoglobin C. For example, people with sickle-hemoglobin C (HbSC) disease have hemoglobin molecules with hemoglobin S and hemoglobin C instead of beta-globin. If mutations that produce hemoglobin S and beta thalassemia occur together, individuals have hemoglobin S-beta thalassemia (HbSBetaThal) disease.

Abnormal versions of beta-globin can distort red blood cells into a sickle shape. The sickle-shaped red blood cells die prematurely, which can lead to anemia. Sometimes the inflexible, sickle-shaped cells get stuck in small blood vessels and can cause serious medical complications.

Related Information

What is a gene?

What is a gene mutation and how do mutations occur?

How can gene mutations affect health and development?

More about Mutations and Health



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Sickle Cell Disease

Also known as Sickle Cell Anemia, Hemoglobin S Leer en español

Sickle cell disease is a group of inherited red blood cells disorde are of African ancestry, but the condition is also common in pec

Early signs and symptoms of sickle cell disease include swelling complications such as infections, delayed growth, and episodes chronic, ongoing pain. Over a lifetime, sickle cell disease can ha

A blood and bone marrow transplant is currently the only cure reduce symptoms and prolong life. Early diagnosis and regular widely from person to person.

Explore this Health Topic to learn more about sickle cell disease













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Causes

Risk Factors

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Information for Health Professionals

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December 03, 2018 | Media Availability

NIH researcher presents encouraging results for gene therapy for severe sickle cell disease

WHAT: A scientist from the National Institutes of Health will present promising, early results from a human...

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List of FDA Orphan Drugs

Sickle cell anemia

Other Names: HbS disease; Hemoglobin S Disease; Sickling disorder due to h Categories: Blood Diseases; Congenital and Genetic Diseases; Endocrine Dise

This disease is grouped under: Hemoglobinopathy

Summary



Sickle cell anemia is a disease in which the body produces abnormally sh crescent or sickle shape. These cells do not last as long as normal, round anemia (low number of red blood cells). The sickle cells also get stuck in Signs and symptoms of sickle cell disease usually begin in early childhoo infections, and periodic episodes of pain (called crises). This condition is and is inherited in an autosomal recessive pattern. [2] Treatment typically may include pain medicines during crises; hydroxyurea to reduce the num vaccines to prevent bacterial infections; and blood transfusions. [1] On Jul approved the use of Endari (prescription grade L-glutamine) to reduce the the first FDA approved treatment that is also available for children with si older.[3]

Symptoms

Treatment



The resources below provide information about treatment options for this condition. If you have questions about which treatment is right for you, talk to your healthcare professional

Management Guidelines

 Project OrphanAnesthesia is a project whose aim is to create peer-reviewed, readily accessible guidelines for patients with rare diseases and for the anesthesiologists caring for them. The project is a collaborative effort of the German Society of Anesthesiology and Intensive Care, Orphanet, the European Society of Pediatric Anesthesia, anesthetists and rare disease experts with the aim to contribute to patient safety.

FDA-Approved Treatments

The medication(s) listed below have been approved by the Food and Drug Administration (FDA) as orphan products for treatment of this condition. Learn more orphan products.

- Hvdroxvurea (Brand name: Droxia) Manufactured by Bristol-Myers Squibb Co FDA-approved indication: To reduce the frequency of painful crises and to reduce the need for blood transfusions in adult patients with sickle cell anemia with recurrent moderate to severe painful crises (generally at least 3 during the preceding 12 months).
- National Library of Medicine Drug Information Portal Medline Plus Health Information
- L-glutamine oral powder (prescription grade) (Brand name: Endari) Manufactured by Emmaus Medical, Inc. FDA-approved indication: To reduce the acute complications of sickle cell disease in adult and pediatric patients 5 years of age and older.
- National Library of Medicine Drug Information Portal
- Hydroxyurea (Brand name: Siklos) Manufactured by Addmedica Laboratories FDA-approved indication: To reduce the frequency of painful crises and to reduce the need for blood transfusions in pediatric patients, 2 years of age and older, with sickle cell anemia with recurrent moderate to severe painful crisis.

National Library of Medicine Drug Information Portal Medline Plus Health Information

Find a Specialist



If you need medical advice, you can look for doctors or other healthcare professionals who have experience with this disease. You may find these specialists through advocacy organizations, clinical trials, or articles published in medical journals. You may also want to contact a university or tertiary medical center in your area, because these centers tend to see more complex cases and have the latest technology and treatments.

Sickle Cell Disease-





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for PATIENT
ORGANIZATIONS

for **INDUSTRY**

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Rare Disease Database

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Sickle Cell Disease

NORD gratefully acknowledges MA Bender, MD, PhD, Department of Pediatrics, University of Washington, Fred Hutchinson Cancer Research Center, Seattle, Washington, for assistance in the preparation of this report.

Synonyms of Sickle Cell Disease

SCD

Subdivisions of Sickle Cell Disease

- sickle cell anemia
- sickle cell hemoglobin C disease
- · sickle cell thalassemia disease

General Discussion

Summary Sickle cell disease (SCD) is a rare blood disorder that is inherited in an autosomal recessive manner. It is characterized by the presence of sickle, or crescent-shaped, red blood cells (erythrocytes) in the bloodstream. These

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Enter a disease name or synonym to search NORD's

Addressing Specific Diagnosis

Genetic testing: prenatal, newborn, child



NIH) U.S. National Library of Medicine



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Genetic Testing

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Summarv

Summary

Genetic tests are tests on blood and other tiss available. Doctors use genetic tests for severa

- Finding genetic diseases in unborn babi
- · Finding out if people carry a gene for a
- · Screening embryos for disease
- Testing for genetic diseases in adults be
- Making a diagnosis in a person who has
- Figuring out the type or dose of a medic

People have many different reasons for being know whether a disease can be prevented or

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Birth Defects

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Research

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Summary

A birth defect is a problem that happens while a baby is developing defects happen during the first 3 months of pregnancy. One out of is born with a birth defect.

A birth defect may affect how the body looks, works or both. Some tube defects are structural problems that can be easy to see. To t use special tests. Birth defects can range from mild to severe. Ca

- Genetics
- Exposures to medicines or chemicals. For example, alcohol syndrome.
- Infections during pregnancy
- · Certain medicines. Before you get pregnant, talk to your he medicines you take.
- · Not getting enough of certain nutrients. For example, not ge during pregnancy is a key factor in causing neural tube def

treatment. But test results might help a person make me decisions, such as ranning pranning or

insurance coverage. A genetic counselor can provide information about the pros and cons of testing.

NIH: National Human Genome Research Institute

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Summary

Genes are the building blocks of heredit instructions for making proteins. Protein place to another, build structures, break

Sometimes there is a mutation, a chang instructions for making a protein, so the cause a medical condition called a genet

You can inherit a gene mutation from one lifetime.

There are three types of genetic disorder

 Single-gene disorders, where a mu Genetic testing and your cancer

Prenatal Testing

Basics	Learn More	See, Play and Learn
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Summary

Prenatal testing provides information about your baby's health before he or she is born. Some routine tests during pregnancy also check on your health. At your first prenatal visit, your health care provider will test for a number of things, including problems with your blood, signs of infections, and whether you are immune to rubella (German measles) and chickenpox.

Throughout your pregnancy, your health care provider may suggest a number of other tests, too. Some tests are suggested for all women, such as screenings for gestational diabetes, Down syndrome, and HIV. Other tests might be offered based on your

- · Personal or family medical history
- · Ethnic background
- · Results of routine tests

There are two types of tests:

 Screening tests are tests that are done to see if you or your baby might have certain problems. They evaluate risk, but do not diagnose problems. If your screening test result is abnormal, it does not mean that there is a problem. It means that more information is needed. Your health



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Alpha fetoprotein

Amniocentesis

Amniocentesis - slideshow

Chorionic villus sampling

Mayo Clinic- prenatal testing



Healthy Lifestyle

Pregnancy week by week

Prenatal testing, including screening and diagnostic tests, can provide valuable information about your baby's health. Understand the risks and benefits.

By Mayo Clinic Staff

Pregnancy is a time of great anticipation — and, sometimes, anxiety. You might worry that your baby will have health problems. While most babies are born healthy, it's important to understand your options for obtaining details about your baby's health.

The two main types of prenatal testing are:

- Screening tests. Prenatal screening tests can identify whether your baby is more or less likely to have certain birth defects, many of which are genetic disorders. These tests include blood tests, a specific type of ultrasound and prenatal cell-free DNA screening. Prenatal screening tests are usually offered during the first or second trimester. Screening tests can't make a definitive diagnosis. If results indicate an increased risk for a genetic disorder, your health care provider will discuss your options for a diagnostic test to confirm the diagnosis.
- Diagnostic tests. If a screening test indicates a possible problem or your age, family history or medical history puts you at increased risk of having a baby with a genetic problem you might consider an invasive prenatal diagnostic test. A diagnostic test is the only way to be sure of a diagnosis. Some diagnostic tests, such as chorionic villus sampling and amniocentesis, carry a slight risk of miscarriage.

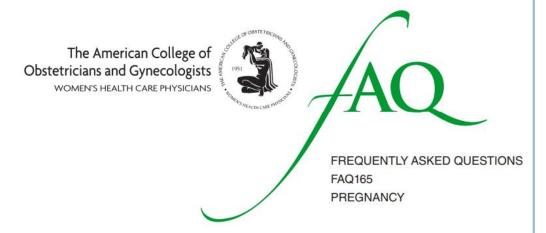
Prenatal screening tests include:

- First trimester screening tests. During your first trimester, your health care provider will offer a blood test and an ultrasound to measure the size of the clear space in the tissue at the back of a baby's neck (nuchal translucency). In Down syndrome and in certain other conditions, the nuchal translucency measurement is abnormally large.
- Second trimester screening tests. During your second trimester, your health care provider will offer another blood test called the quad screen. This test measures levels of four substances in your blood. Results indicate your risk of carrying a baby who has certain chromosomal conditions, such as Down syndrome. The test can also help detect neural tube defects serious abnormalities of the brain or spinal cord.
- Prenatal cell-free DNA screening. This blood test examines fetal DNA in the maternal bloodstream to screen for the increased chance for specific chromosome problems, such as Down syndrome. This screening can also provide information about a baby's sex and rhesus (Rh) blood type.

Prenatal screening tests for fetal abnormalities are optional. It's important to make an informed decision about prenatal testing, especially if you're screening for fetal conditions that can't be treated. Before going forward, consider these guestions:

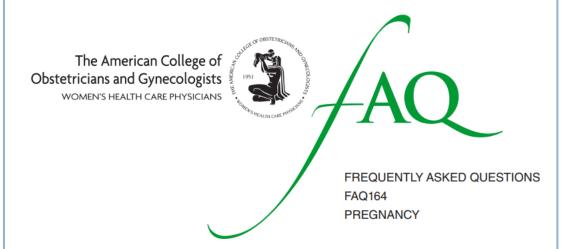
- What will you do with the test results? Normal results can ease your anxiety. However, if prenatal testing indicates that your baby might have a birth defect, you could be faced with wrenching decisions such as whether to continue the pregnancy. On the other hand, you might welcome the opportunity to plan for your baby's care in advance.
- Will the information shape your prenatal care? Some prenatal tests detect problems that can be treated during pregnancy. In other cases, prenatal testing alerts your health care provider to a condition that requires immediate treatment after birth.
- . How accurate are the results? Prenatal screening isn't perfect. The rate of inaccurate results, known as false-negative or false-positive results, varies from test to test.
- What are the risks? Weigh the risks of specific prenatal tests such as anxiety, pain or possible miscarriage against the value of knowing the results.

ACOG- prenatal genetic testing



Prenatal Genetic Screening Tests

- . What is prenatal genetic testing?
- · What are genetic disorders?
- . What are the two main types of prenatal genetic tests?
- · What are the different types of prenatal genetic screening tests?
- . What is first-trimester screening?
- What is second-trimester screening?
- · What is combined first- and second-trimester screening?
- What is cell-free DNA testing?
- · What do the different results of prenatal screening tests mean?
- · How accurate are prenatal genetic screening tests?
- . What should I consider when deciding whether to have prenatal genetic testing?
- Glossary



Prenatal Genetic Diagnostic Tests

- What is prenatal genetic testing?
- · What are genetic disorders?
- What are the two main types of prenatal genetic tests?
- What is amniocentesis?
- . What is chorionic villus sampling?
- · What is preimplantation genetic diagnosis?
- How are the cells analyzed in prenatal diagnostic testing?
- What do the different results of prenatal diagnostic tests mean?
- What should I consider when deciding whether to have prenatal genetic testing?
- How do I choose between prenatal screening and diagnostic testing?
- Glossary



Newborn Screening Welcome

SCREENING PROGRAMS

The National Newborn Screening and Global Reso independent U.S. national resource center for new screening information globally. It serves as the only of newborn screening information. Its director is cu Pediatrics at the University of Texas Health Science adjunct capacity. The NNSGRC was originally four US Health Resources and Services Administration January 1, 2013.

Originally created as a federal focal point for nation in the US, the NNSGRC is dedicated to providing resource information both nationally and globally. consultative services, program reviews, selected n international discussion listsery, the NNSGRC probenefit national and international newborn screeni

- Health Professionals
- Consumers
- The Public Health Community
- US and Other Government Officials
- Healthcare and Other Policy Makers
- Non-Government Organizations

NENSGRC National Newborn Screening & Global Resource Center

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FAMILIES

Educational Materials

SCREENING PROGRAMS

"The Story of Newborn Screening" by Harvey Levy, MD:

GENERAL INFORMATION

A 10-part webcast from the New England Regional Consortium describing the viewed in its entirety or in individual segments).

Newborn Screening Education Brochures

Brochure for Parents - Model developed for state use based on parent focus d Brochure for Providers: - Model developed for state use based on provider for **Brochure for Grandparents**

and Translation rating system

Disease Specific Brochures

Samples of patient education brochules*. Click on a state/program abbreviation languages listed as available.

3-MCC Deficiency

English: KY

5-Oxoprolinuria

English: IL

Alpha Thalassemia

English: WA

Cambodian: WA

Chinese: WA

Laotian WA

Spanish WA

Vietnamese WA

Arginosuccinic Acidemia

English KY

For Families

Frequently Asked Questions about Newborn Bloodspot Screening

Find out the answers to the following questions and more. Why does my baby need newborn screening tests? How will my baby be tested? How will I get the results of the test?

Conditions Required or Otherwise Included in State Newborn Screening Programs

A comprehensive listing (in chart format) that shows the implementation status of the various conditions on the nationally Recommended Uniform Screening Panel (RUSP). The current RUSP includes 31 conditions. Other conditions may be required by some states and these are also included. Readers should be sure to understand which conditions are required by state law or rule and those that may not be required but may be included as a result of the technology being used. Notes at the top of the table explain the symbols used in the tables.

State Newborn Screening Program Contacts

A PDF file containing contact information for the laboratory and follow-up coordinators in each state's and territory's newborn screening program.

Hearing Screening

Hearing screening resources, state screening programs, legislation, organizations, and parent education links.

Disorder Fact Sheets

Disorder Fact Sheets for parents provided by the Screening, Technology And Research in Genetics (STAR-G) Project.

Additional Testing

List of screening laboratories that may charge a fee for additional screening not offered in some screening programs.

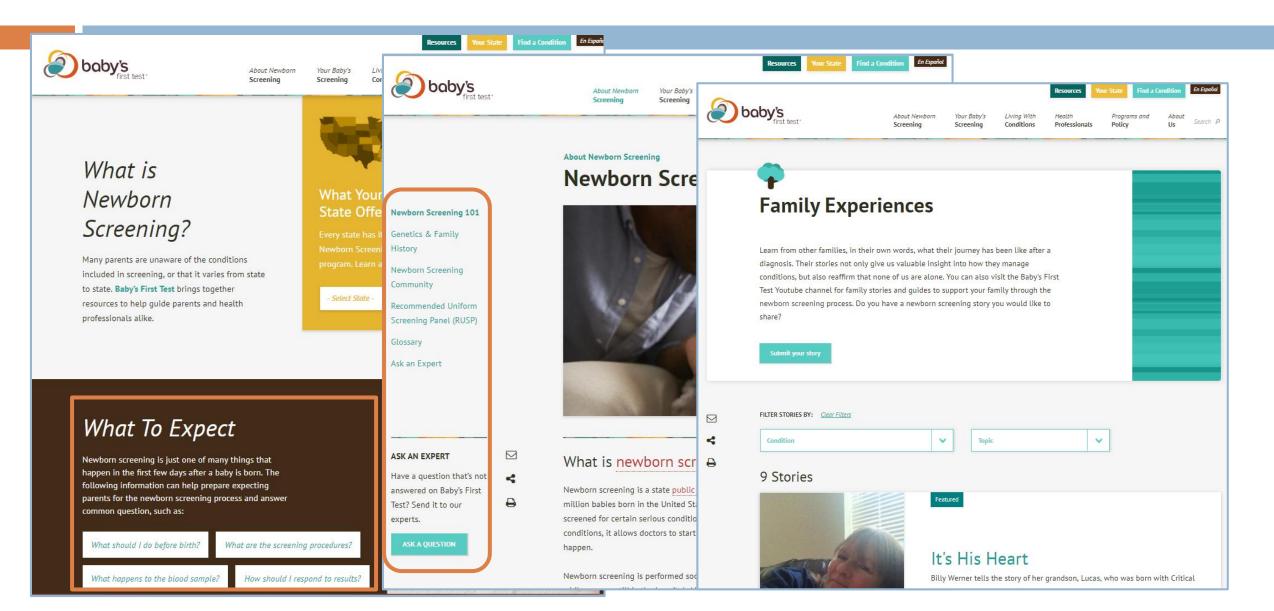
Genetics Information

Links to genetics counseling, family forums, and fact sheets on genetics.

Support and **Advocacy Groups** Links to support groups for newborn screening disorders,



Baby's First Test



About Genetic Counselors

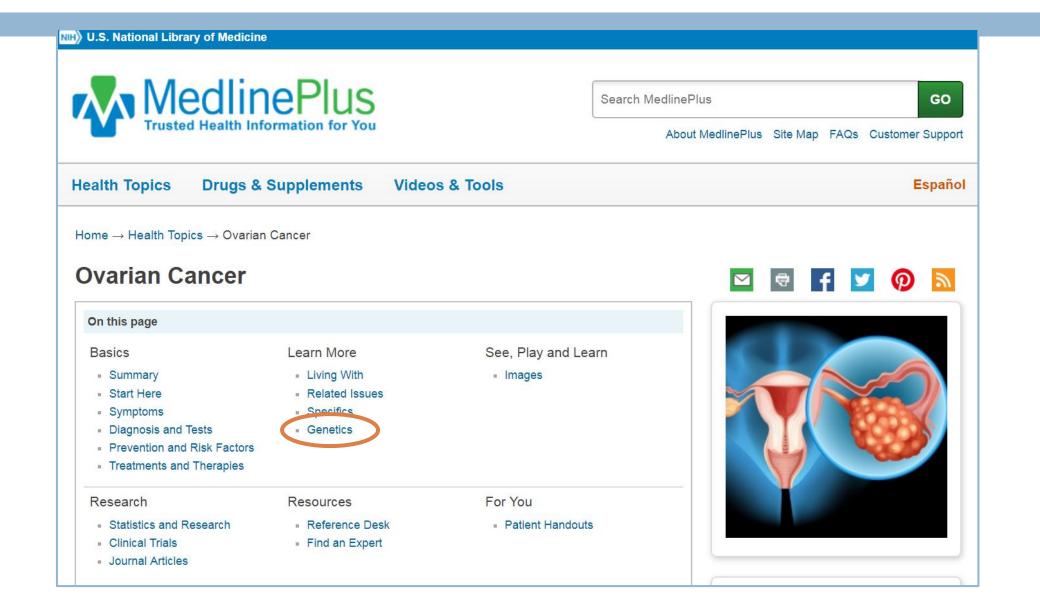


About Genetic Counselors - prenatal genetic counseling

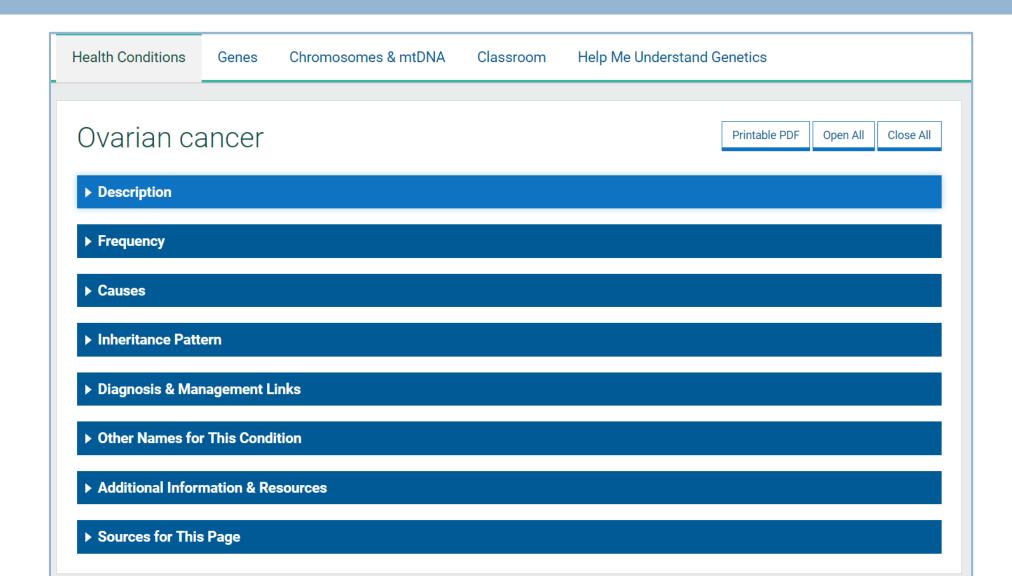
Addressing Specific Diagnosis

Ovarian Cancer

MedlinePlus- Ovarian Cancer



Genetics Home Reference- Ovarian Cancer



ABOUT CANCER

CANCER TYPES

Home > Cancer Types

Ovarian, Fallopian Tube, Cancer-Patient Version

OVERVIEW

Ovarian epithelial cancer, fallopian tube canc cancer form in the same kind of tissue and ar These cancers are often advanced at diagnos ovarian tumors include ovarian germ cell tun malignant potential tumors. Explore the links about treatment, prevention, screening, rese these conditions.

Select a cancer type to view general and information

CAUSES & PREVENTION

PDO Prevention Information for Patie

Ovarian, Fallopian Tube, & Primary Peritone

More information

BRCA Mutations: Cancer Risk and Genetic

STATISTICS

Ovarian Cancer | Did You Know?





NIH > NATIONAL CANCER INSTITUTE

Español

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CANCER TYPES

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Home > Cancer Types > Ovarian, Fallopian Tube, and Primary Peritoneal Cancer > Patient

CANCER CAUSES AND PREVENTION

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NCI Cancer Genetics Services Directory

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OVARIAN. **FALLOPIAN TUBE. AND PRIMARY** ON **PERITONEAL** CANCER

Patient

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Ovarian Epithelial. Fallopian Tube, & Primary Peritoneal Cancer Symptoms, Tests, Prognosis, & Stages

Treatment of Ovarian Epithelial, Fallopian Tube. & Primary Peritoneal Cancer

Ovarian Germ Cell Tumors Symptoms, Tests, Prognosis, & Stages

Treatment of Ovarian Germ Cell Tumors

Ovarian Low Malignant Potential Tumors Symptoms, Tests, Prognosis, & Stages

Treatment of Ovarian Low Malignant Potential Tumors

Prevention of Ovarian, Fallopian Tube, & Primary Peritoneal Cancer

Screening for Ovarian, Fallopian Tube, & Primary **Peritoneal Cancer**

Ovarian, Fallopian Tube, and Primary Peritoneal Cancer Screening (PDQ®)-Patient Version

Go to Health Professional Version (



ON THIS PAGE

- What is screening?
- General Information About Ovarian, Fallopian Tube, and Primary Peritoneal Cancer
- Ovarian, Fallopian Tube, and Primary Peritoneal Cancer Screening
- Risks of Ovarian, Fallopian Tube, and Primary Peritoneal Cancer Screening
- About This PDQ Summary

What is screening?

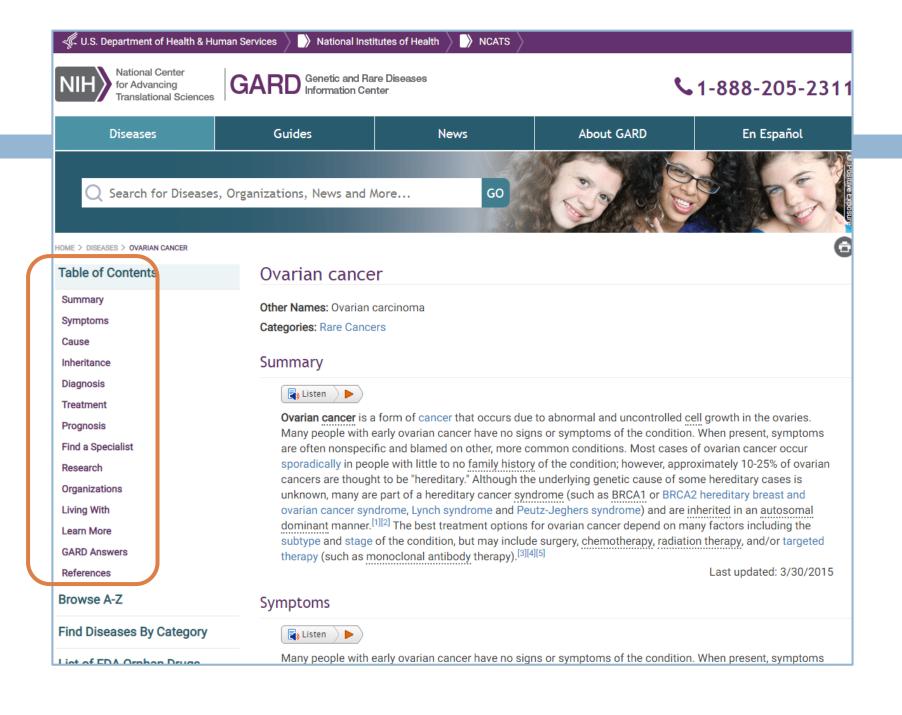
Screening is looking for cancer before a person has any symptoms. This can help find cancer at an early stage. When abnormal tissue or cancer is found early, it may be easier to treat. By the time symptoms appear, cancer may have begun to spread.

Scientists are trying to better understand which people are more likely to get certain types of cancer. They also study the things we do and the things around us to see if they cause cancer. This information helps doctors recommend who should be screened for cancer, which screening tests should be used, and how often the tests should be done.

It is important to remember that your doctor does not necessarily think you have cancer if he or she suggests a screening test. Screening tests are given when you have no cancer symptoms.

If a screening test result is abnormal, you may need to have more tests done to find out if you have cancer. These are called diagnostic tests.

Specific inherited mutations in BRCA1 and BRCA2 most notably increase the risk of female breast and ovarian cancers, but they have also been associated with increased risks of several additional types of cancer. People



What You Need to About Ovarian Cand

Basic Information



Learn how to lower your risk and about the symptoms, risk factors, and treatment for ovarian cancer.

Statistics



Ovarian can than any oti Data Visuali statistics.

Knowledge Is Power: Ovarian Cancer

OVARIAN CANCER

There are five main types of cancer that affect a woman's reproductive organs: cervical, ovarian, uterine, vaginal, and vulvar. As a group, they are referred to as gynecologic (GY-neh-kuh-LAH-jik) cancer. (A sixth type of gynecologic cancer is the very rare fallopian tube cancer.)

This fact sheet about ovarian cancer is part of the Centers for Disease Control and Prevention's (CDC) Inside Knowledge: About Gynecologic Cancer campaign. The campaign helps women get the facts about gynecologic cancer, providing important "inside knowledge" about their bodies and health.

What is ovarian cancer?

Cancer is a disease in which cells in the body grow out of control. Cancer is always named for the part of the body where it starts, even if it spreads to other body parts later.

When cancer starts in the ovaries. it is called ovarian cancer. Women have two ovaries that are located in the pelvis, one on each side of the uterus. The ovaries make female hormones and produce eggs.

When ovarian cancer is found in its early stages, treatment is most effective.

Who gets ovaria

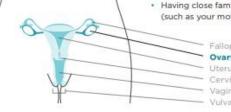
All women are at ris likely to get the diswomen who get over number of ovarian

Each year, approxin cancer. Among wor most common cand

What raises a wo of getting ovariar

There is no way to kr you will get ovarian women who get it do history of ovarian ca the following factor a woman's risk for o

. Being middle-age



Inside Knowledge About Gynecologic Cancer

Inside Knowledge Campaign Print Materials Public Service Announcements and Videos Survivor Stories Campaign Research Provider Continuing Education Buttons and Badges About the Campaign

Test Your Inside Knowledge Stay Informed







CDC > Cancer Home > Gynecologic Cancers

Inside Knowledge About Gynecologic Cancer





Language: English (US)



The Inside Knowledge campaign raises awareness of the five main types of gynecologic cancer: cervical, ovarian, uterine, vaginal, and vulvar, Inside Knowledge encourages women to pay attention to their bodies, so they can recognize any warning signs and seek medical care. Inside Knowledge offers a variety of television and radio public service announcements in English and Spanish, as well as other videos and educational resources for women and health care providers.

Posters with Cote de Pablo



Videos, PSAs, Posters, and More!





"Are You Listening?" TV and Radio PSAs

Additional Resources

Inside Knowledge provides resources to women, for health care providers, and organizations to share with patients and communities.

- NEW! Shareable buttons and badges.
- NEW! Short animated videos
- PSAs
- Posters
- · Information about each of the most common gynecologic cancers: cervical, ovarian, uterine, vaginal, and vulvar
- . NEW! Family History and Cancer Fact Sheet Printable version 7 [PDF-12MB]
- · Provider continuing education (CE) on gynecologic cancers
- · Information about how and why Inside Knowledge was developed

cdc.gov/cancer/knowledge

800-CDC-INFO

Health and Wellness





What can we help you find?

Diseases and Conditions

Call the OWH HELPLINE: 1-800-994-9662 9 a.m. - 6 p

Patient Materials

Reproductive Health

A FACT SHEET FROM THE OFFICE ON WOMEN'S HEALTH



Home > Cancer > Ovarian cancer

Cancer

Breast cancer

Cervical cancer

Ovarian cancer

Uterine cancer

View A-Z health topics



Ovarian cancer fact sheet (PDF, 182 KB)



Ovarian cancer

Ovarian cancer is cancer that begins in the ovaries. The ovaries make female hormones and produce a woman's eggs. Ovarian cancer is a serious cancer that is more common in older women. Treatment is most effective when the cancer is found early. Screening for ovarian cancer is not recommended for most women.

Learn more about ovarian cancer at the National Cancer Institute.

Expand a

What is ovarian cancer?

Who gets ovarian cancer?

Are some women more at risk for ovarian cancer?

Women with a high risk of ovarian cancer are those with a harmful mutation on the BRCA1 or

Ovarian Cancer

Ovarian cancer is cancer that begins in the ovaries. The ovaries make female hormones and produce a woman's eggs. Ovarian cancer is a serious cancer that is more common in older women. Treatment is most effective when the cancer is found early.

Q: What is ovarian cancer?

A: Ovarian cancer forms in tissues of the ovary. (An ovary is one of a pair of female reproductive glands in which the ova, or eggs, are formed.)

Tumors in the ovaries can be benign, which means they are not cancer, or they can be malignant, which means they are cancer.

Cancers that start in the ovaries can spread to other parts of the body. This is called metastasis. Cancer that starts in the ovaries and spreads to other parts of the body is still called ovarian cancer.

Q: Who gets ovarian cancer?

A: Around one in every 60 women in the United States will develop ovarian cancer. Most ovarian cancers are diagnosed in women over 60, but this disease can also affect younger women. Among women in the United States, ovarian cancer is the eighth most common cancer and the fifth leading cause of cancer death.

Q: Are some women more at risk for ovarian cancer?

A: Women with a high risk of ovarian cancer are those with a harmful mutation on the BRCA1 or BRCA2 genes. These mutations can be found with a blood test. Women with a family or personal history

of breast or ovarian cancer also have a higher risk of ovarian cancer.

If you have family members in multiple generations with breast cancer or ovarian cancer, see your doctor to learn more about your risk of ovarian cancer. Research shows that certain steps, such as surgery to remove the ovaries and the fallopian tubes, may help prevent ovarian cancer in women who are at high risk. The sooner ovarian cancer is found and treated, the better your chance for recovery. But ovarian cancer is hard to detect early because its symptoms are also the symptoms of many other illnesses.

Q: What are the symptoms of ovarian cancer?

A: The following may be symptoms of ovarian cancer if they continue or get worse over time:

- · Pain in the pelvis or abdomen (belly)
- · Bloating in the abdomen
- Urinary urgency (needing to pee right away)
- · Urinary frequency (having to pee often)
- · Constipation or diarrhea
- · Feeling full quickly while eating
- · Having difficulty eating
- Vaginal bleeding or other discharge that is different than normal
- · Back pain

If you have any of these symptoms, talk to your doctor. He or she can determine if the cause is cancer or







Español

1-800-4-CANCER Live Chat **Publications** Dictionary

ABOUT CANCER

CANCER TYPES

RESEARCH

GRANTS & TRAINING

NEWS & EVENTS

ABOUT NCI

search

Q

Home > About Cancer > Cancer Causes and Prevention









CANCER CAUSES AND PREVENTION

Risk Factors

Genetics

NCI Cancer Genetics Services Directory

Cancer Prevention Overview

Research

The Genetics of Cancer

ON THIS PAGE

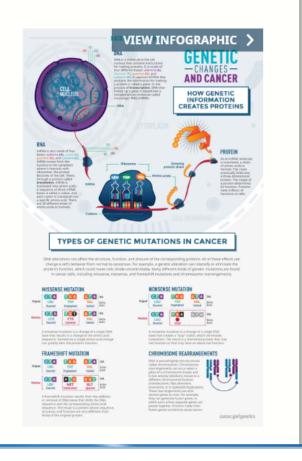
- Genetic Changes and Cancer
- Hereditary Cancer Syndromes
- Genetic Tests for Hereditary Cancer Syndromes
- Identifying Genetic Changes in Cancer

Genetic Changes and Cancer

Cancer is a genetic disease—that is, cancer is caused by certain changes to genes that control the way our cells function, especially how they grow and divide.

Genes carry the instructions to make proteins, which do much of the work in our cells. Certain gene changes can cause cells to evade normal growth controls and become cancer. For example, some cancer-causing gene changes increase production of a protein that makes cells grow. Others result in the production of a misshapen, and therefore nonfunctional, form of a protein that normally repairs cellular damage.

Genetic changes that promote cancer can be inherited from our parents if the changes are present in germ cells, which



Whether you work in a hospital, public, or health sciences library you may have a patron or patient seeking information about a particular health condition such as Alzheimer's disease either for themselves or someone they know. As genetics becomes more commonly referred to in relation to Alzheimer's it is good to be able to have a variety of resources available to use or to refer others to as patients may request information about their own risk or the risk for their children. Using the resources mentioned in this class see if you can locate information for the following questions.

It is recommended you start with Genetics Home Reference and from there explore the other resources mentioned in this class

Please state which resource you used to answer each question

- □ What genes are associated with early onset Alzheimer's?
- What genes are associated with late onset Alzheimer's?
- What resource would you use that provides the patron with information about how Alzheimer's runs in the family?
- What resource would you offer a patron to learn more about the basics of genetics?

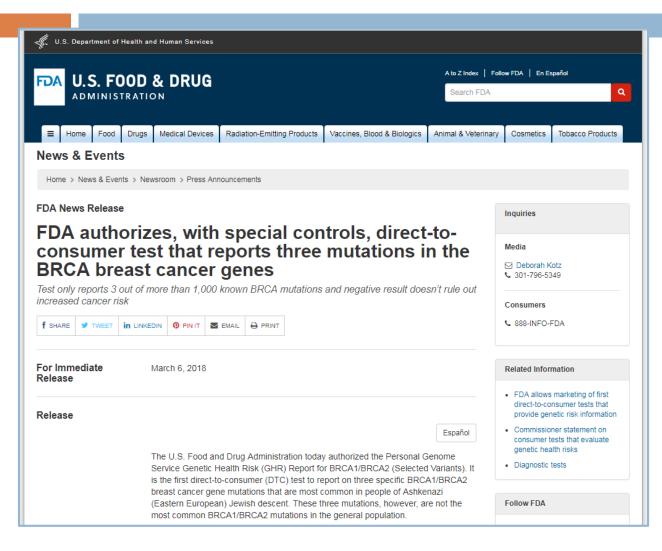
Direct to Consumer Genetic Testing

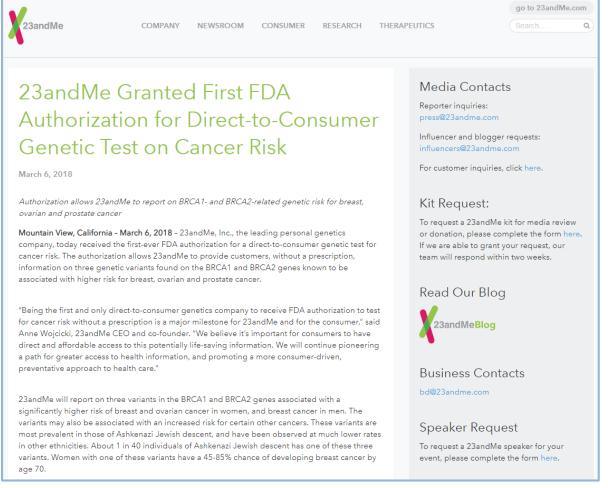
Direct to Consumer Genetic Testing Companies





DTC BRCA test





FDA announcement

23andMe announcement

Direct a revised posit of M

Disclaimer: These recommendations are de cational resource for medical geneticists ar ers to help them provide quality medical ger these recommendations does not necessarily outcome. These recommendations should n all proper procedures and tests or exclusive that are reasonably directed to obtaining th ing the propriety of any specific procedure

With ongoing genetic discoveries and nology, more genetic tests are available with greater availability has come incre for genetic tests and expansion of dire The American College of Medical C (ACMG) has revised its 2008 e-publica (ACMG Statement on Direct-to-Conretired; available by request to acmg@ that it is critical for the public to realize only one part of a complex process th assessment, diagnosis, and disease r

results of such testing can have important meanin implications for individuals and their family members. ACMG believes that the following should be considered the minimum requirements Consensus statement



Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement

Nick Webborn, ¹ Alun Williams, ² Mike McNamee, ³ Claude Bouchard, ⁴ Yannis Pitsiladis, ⁵ Ildus Ahmetov, ⁶ Euan Ashley, ⁷ Nuala Byrne, ⁸ Silvia Camporesi, ⁹ Malcolm Collins, ¹⁰ Paul Dijkstra, ¹¹ Nir Eynon, ¹² Noriyuki Fuku, ¹³ Fleur C Garton, ¹⁴ Nils Hoppe, ¹⁵ Søren Holm, ¹⁶ Jane Kaye, ¹⁷ Vassilis Klissouras, ¹⁸ Alejandro Lucia, ¹⁹ Kamiel Maase, ²⁰ Colin Moran, ²¹ Kathryn N North, ¹⁴ Fabio Pigozzi, ²² Guan Wang⁵

Position Statem

Purpose

This position statemer policymakers and the decision-making about (DTC-GT). The focus of such as disease risk as conditions. It does not wellness and trait char knowledge and policies relevant to future dire

Background

Direct-to-consumer ge genetic/genomic tests testing that detects va genome panels (e.g., broad-based risk asses

Within any clinical hea aim to protect patients DTC-GT, varies accordi Ianuale, Ricciardi & Bd

 Additional material is published online only. To view please visit the journal online (http://dx.doi.org/10.1136/ bjsports-2015-095343).

For numbered affiliations see end of article.

Correspondence to

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Accepted 25 September 2015

ABSTRACT

The general consensus among sport and exercise genetics researchers is that genetic tests have no role to play in talent identification or the individualised prescription of training to maximise performance. Despite the lack of evidence, recent years have witnessed the rise of an emerging market of direct-toconsumer marketing (DTC) tests that claim to be able to identify children's athletic talents. Targeted consumers include mainly coaches and parents. There is concern among the scientific community that the current level of knowledge is being misrepresented for commercial purposes. There remains a lack of universally accepted guidelines and legislation for DTC testing in relation to all forms of genetic testing and not just for talent identification. There is concern over the lack of clarity of information over which specific genes or variants are being tested and the almost universal lack of appropriate genetic counselling for the interpretation of the genetic data to consumers. Furthermore independent studies have identified issues relating to quality control by DTC wrong lest, and mismerpretation of results leading to different results being reported from

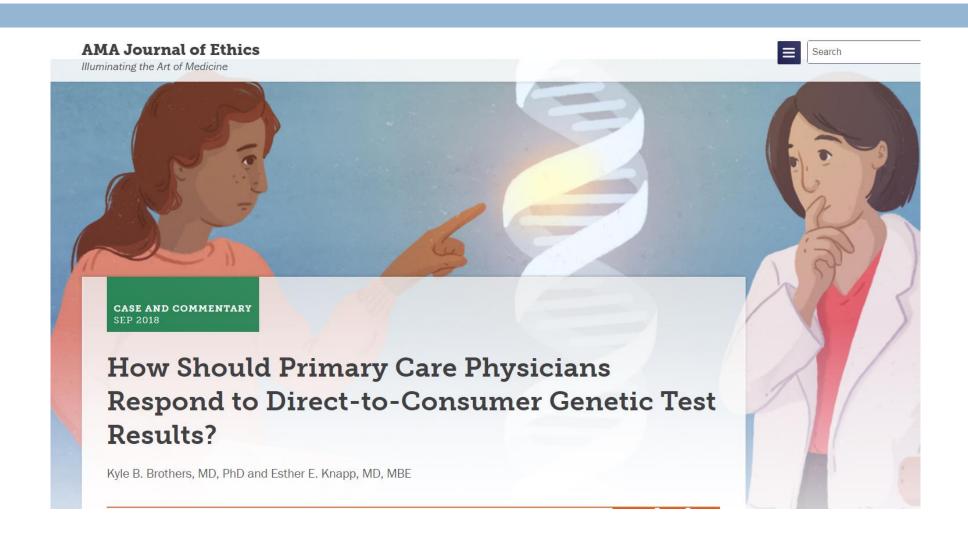
of the evidence in relation to genetic testing and the limitations of current knowledge. This article reviews the issues around the currently available evidence behind the genetic testing, comments on the ethical considerations and makes recommendations about such tests.

STATEMENT ON BACKGROUND TO THE **CONSENSUS PROCESS**

A group of world experts in the field of genomics, exercise, sport performance, disease, injury and antidoping gathered with the International Federation of Sports Medicine (FIMS) Scientific Commission for a symposium to discuss the current state of knowledge and to share ideas. One key concern was the misuse of research evidence and the misinformation about genetic testing, particularly when marketed directly to the public, coaches or parents. This is known as DTC testing for the purpose of talent identification and to assess potential for future sports performance. There have been

inappropriate choices for disease management or preven-

AMA commentary for physicians



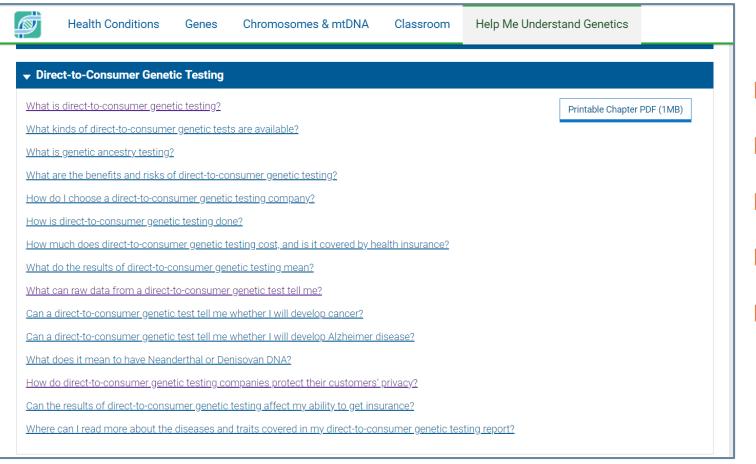
Concerns

- Privacy
- Legality
- Who has access?
- ☐ How useful now?
- What all is being done now and in the future with the information?
- Unexpected surprises?
- □ Test results can vary among companies
- Validity of tests
- No counseling provided
- Who can get the testing?

Benefits

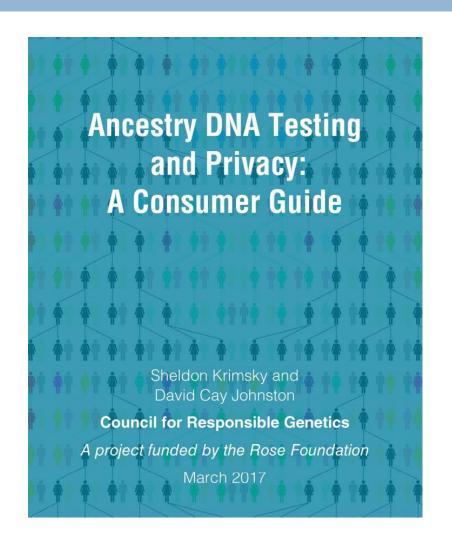
- Learn more about own health
- More effective medical treatments
- Learn more about ethnicity and family history
- Bring awareness to family health issues for future generations
- Motivation to work on health habits
- Encourages patient engagement
- Contributing to advancement of healthcare and science
- Moral obligation

DTC- Genetics Home Reference



- What kinds are available
- Ancestry testing
- What do results mean
- Risk of developing a disease
- Privacy protection

Information for Patients/Consumers about DTC





What is direct-to-consumer (DTC) genetic testing?

DTC refers to a genetic test you can complete at home without a health care provider. You collect a DNA sample and send it to the company. They analyze it and give you a report on your genetics

What information can I get from a DTC genetic test?

There are many different types of DTC tests available. Some tell you about ancestry, kinship, lifestyle factors and disease risk. Companies can analyze your DNA and give information about these things.

What do DTC companies do with my data?

That depends on the fine print! You should read up on each company's policies. They vary on how they decide to store your sample and your data, and with



Genetic Counselors

Personalized Care For Your Genetic Health



ABOUT GENETIC COUNSELORS GENETIC COUNSELING GENETIC TESTING

GENETIC CONDITIONS FAOS + RESOURCES

Home >> Genetic Testing >> What is At-Home Genetic Testing?

What is At-Home Genetic Testing?

Understanding what an at-home test will - and will not - tell you

For a price (typically several hundred to a thousand dollars) you can order a genetic testing kit online or by phone. You'll swab your cheek or spit into a test tube. Then you will mail it to a lab where it may be tested for a wide variety of things - from whether you inherited your intolerance to the lactose in dairy products to your risk of certain types of cancer to if you carry a gene for a serious illness such as Cystic Fibrosis and could pass it on to your children.

There are at-home tests for:

- Traits (e.g., male hair loss to dimples)
- Wellness (e.g., risk of certain types of cancer to restless leg syndrome)
- · Ancestry reports (i.e., ethnicity and lineage)
- · Carrier status (e.g., Tay-Sachs Disease to Sickle Cell Anemia)
- Paternity testing (i.e., determining a child's biological father)

There are three general ways to get genetic testing:

. Through your physician or a genetic counselor - These are the most detailed and comprehensive tests available. They include cancer testing and testing for genetic disorders. They include large "panels,"

Related Links



FIND A GENETIC COUNSELOR



PRENATAL CONDITIONS



FAMILY HEALTH HISTORY



PROTECTING YOUR PRIVACY



- From the National Society of Genetic Counselors
- Plain language
- Also a guide to at-home BRCA testing
- Glossary of terms

Genetic Counselors

- Evaluate family history and medical records
- Assist in making decisions regarding genetic testing
- Identify and interpret risks of inherited disorders, increase the family's understanding of a genetic condition
- Discuss options regarding disease management and the risks and benefits of further testing and other options
- Help the individual and family identify the psychosocial tools required to cope with potential outcomes
- Reduce the family's anxiety

Deciding about genetic testing







An initiative of the ABIM Foundation

Making smart decisions about genetic testing:

Avoid unneeded tests, and understand the tests you need

- enetic testing can help identify an inherited condition or disease risk. The test results might help you and your doctor:
- Choose ways to prevent or treat a condition.
- Decide which screening tests you need (to find a disease at an early stage when it might be more treatable).

Genetic testing may also tell you which family members are at risk.

But sometimes a genetic test is not the best way to find an inherited condition or disease risk. A routine blood test or procedure might be just as good. And it might be less costly and more easily available.

Know what to expect.

Before you have any genetic test, you should understand its possible benefits, harms, and limitations. And you should think about how it might affect others in your family.

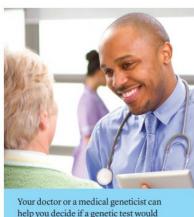
Talk to your doctor or a medical geneticist.

You should discuss the reasons for a genetic test with your doctor before your sample for testing is collected. Your doctor may refer you to a medical geneticist. This is a specialist who is trained to:

· Diagnose genetic conditions.

be useful to you.

- · Select the most appropriate genetic tests.
- · Explain test results to patients and their families.
- Recommend personalized treatment and prevention options.



Questions to ask:

- Am I in the group at risk and should I get tested?
- > If I decide to get tested, what do the results mean?
- What are my treatment options based on results?
- How do I decide on treatment?

Choosing Wisely and genetic testing

My Family Health Portrait

A tool from the Surgeon Genera

Using My Family Health Portrait you can:

- · Enter your family health history.
- · Learn about your risk for conditions that can run in families.
- Print your family health history to share with family or your health care provider
- · Save your family health history so you can update it over time.

Talking with your health care provider about your family health history can help you stay healthy!

Learn more about My Family Health Portrait

Create a Family Health History

Use a Saved History





Knowing is Not Enough—Act on Your Family Health History

Español (Spanish)

Has your mother or sister had breast cancer? Does your mother, father, sister, or brother have diabetes? Did your mother, father, brother, or sister have colorectal (colon) cancer before age 50? If you answered "yes," you are more likely to get the same disease as your parent or sibling and should consider earlier screening. Talk to your doctor about when to start screening and what other steps to take to prevent the disease or find it early. Finding disease early can often mean better health in the long run.



Did you know that Thanksgiving is also National Family History Day? Even if you don't have a parent or sibling with cancer or diabetes, you might be more likely to get a disease if other

people in your family have or had the disease. However, having a family health history of a disease doesn't mean that you will definitely get it. Knowing your family health history risk can help you—if you act on it. Collecting your family health history is an important first step. Whether you know a lot about your family health history or only a little, take time to talk to your family about their health histories at family gatherings this holiday season. It might not be easy. Your family members might not be used to talking about their diseases or might not want to talk. But starting the conversation is important. Remember, you're asking not just for your own health, but for the health of everyone in your family.

How to Collect Your Family Health History

- Talk to your family. Write down the names of your close relatives from both sides of the family: parents, siblings, grandparents, aunts, uncles, nieces, and nephews. Talk to these family members about what conditions they have or had, and at what age the conditions were first diagnosed. You might think you know about all of the conditions in your parents or siblings, but you might find out more information if you ask.
- Ask questions. To find out about your risk for <u>chronic diseases</u>, ask your relatives about which of these diseases they have had and when they were diagnosed. Questions can include:
 - Do you have any chronic diseases, such as heart disease or diabetes, or health conditions, such as high blood pressure or high cholesterol?
 - Have you had any other serious diseases, such as cancer or stroke?
 What type of cancer?
 - How old were you when each of these diseases or health conditions was diagnosed? (If your relative doesn't remember the exact age, knowing the approximate age is still useful.)

Don't know where to start? My Family Health Portrait can

help!

My Family Health Portrait

My Family Health Portrait is a free and easy to use online tool to help you collect your family health history information. You can share your information with your family members and doctor.

- What is your family's ancestry? From what countries or regions did your ancestors come to the United States?
- What were the causes and ages of death for relatives who have died?

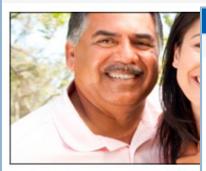
CDC – Family Health History

Family Health History and Chronic Disease

Learn more about what having a family health history of each of these conditions means for you:

Family Health History

Genomics



Knowing and acting on your family health histor

Your doctor can use it to develop a more compl



The Basics



Planning for Pregnancy



During Pregnancy



For Children

Family Health History

Genomics > Family Health History

♠ Family Health History

The Basics

Family Health History & Chronic

Planning for Pregnancy

During Pregnancy

For Children

For Adults

Information for Health Professionals

Information for Researchers

Tools and Resources

Family Heal

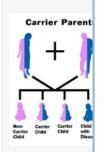
or daddy's dimples. But yo parents' family health hist have a family health histor screening disorder, or gen condition. Knowing if your important so that you can

Other reasons for genetic

- A previous pregnancy

baby could have health iss

Carrier Screening



Expecting a baby? You mig

Based on your family healt

- · Infertility (trouble get
- 2 or more miscarriage
- A baby who died at le

After genetic counseling, y



disease and catch it early if it develops.

Breast & Ovarian Cancer

Colorectal Cancer and Lynch Syndrome

If you have a family health history of a chronic disease like cancer, heart disease, diabetes, or osteoporosis, you are more likely to get that disease yourself. Share your family health history with your doctor, who can help you take steps to prevent



Hereditary Hemochromatosis



Heart Disease, Genetics and Family History



Osteoporosis



racial or ethnic group. Talk to your doctor if you want to know about carrier screening for genetic diseases that run in your family. In addition, current recommendations 1 [7] state that all women should be offered carrier screening for

If the results show that you are a carrier for a disease, the other potential parent would also need to have carrier screening to know if you could have a baby with the diseas

CDC Family Health History

Help

Disclaimer: The Surgeon General's My Family Health Portrait tool does NOT keep a government record of the information you fill in nor make your health information available to anyone else but you. It only provides the software for organizing your information. By accessing the tool on the web, you make use of that software, But the information you fill in is not transmitted back to our servers, and never available to anyone else, unless you choose to share or disclose it. After you fill in

		Disease or Condition	1		
Your Health Information the list below, select a		pdown box. Then select the Age at Diagnosis and press the Add button. You may repeat	this process as necessary.		
Weight	lbs ▼				
Height	Feet Inches -OR-	Centimeters			
Were you adopted?	□ Yes	, ,			
Were you born a twin	?● No ○ Yes - Identical (Same) ○ Ye	_ ' ' ' ' ' ' ' ' ' ' ' ' ' ' ' ' ' ' '			
*Date of Birth		(mm/dd/yyyy)			
*Sex assigned at birth	: Male Female				
*Name:					
We start the family health the health history. • Indicates required infor		nal information and your health history information. At the bottom of the page (you may	need to scroll), press the 'Next' button. You will then be asked to tell th	e system which family members you wou	ld like to add to
Your Personal Inform	nation				
	ion that will be useful for you and yo				

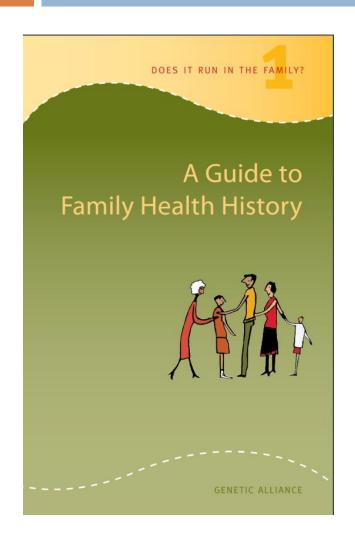
American Indian or Alaska Native
Asian
Black or African-American Race: ■ Native Hawaiian or Other Pacific Islander ■ White

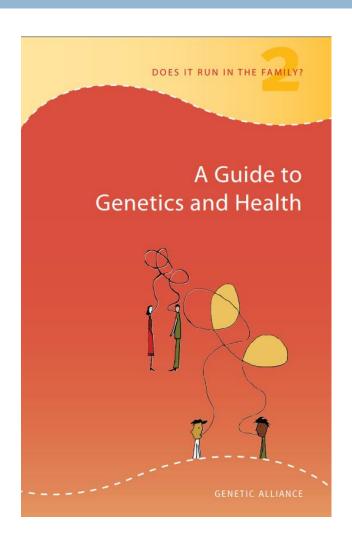
☐ Hispanic or Latino ☐ Ashkenazi Jewish ☐ Not Hispanic or Latino

Why are we asking about Ashkenazi Jewish heritage?

Ethnicity:

Genetic Alliance guides





Ethics and Privacy

Societal Concerns

- Who should have access to personal genetic information, and how will it be used?
- Who owns and controls genetic information?
- How does personal genetic information affect an individual and society's perceptions of that individual?
- □ What are the larger societal issues raised by new reproductive technologies?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- □ How do we prepare healthcare professionals and the public?
- What is considered acceptable diversity?
- Where is the line between medical treatment and enhancement?
- □ Should testing be performed when no treatment is available?

Law Enforcement

AMERICA

In Hunt For Golden State Killer, Investigators Uploaded His DNA To Genealogy Site

by LAUREL WAMSLEY

April 27, 2018 • After failing to find a match within criminal databases, law enforcement uploaded the killer's DNA profile to a no-frills website used to trace ancestry. The tactic has spurred privacy concerns.

GINA



GENETIC INFORMATION NONDISCRIMINATION ACT

About

| Contact

Genetic Information

What is genetic information and why is it important?

GINA & Health Insurance

What are GINA's health insurance protections?

GINA & Employment

What are GINA's employment protections?

What is GINA?

The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects individuals from genetic discrimination in health insurance and employment. Genetic discrimination is the misuse of genetic information. This resource provides an introduction to GINA and its protections in health insurance and employment. It includes answers to common questions and examples to help you learn. Choose from one of the boxes to the left to begin!

- Have questions, comments or suggestions? Send us a note.
- ☐ Click here for a printer friendly version.
- For healthcare provider resources click here.
- ☐ Click here for the GINA & You Information Sheet

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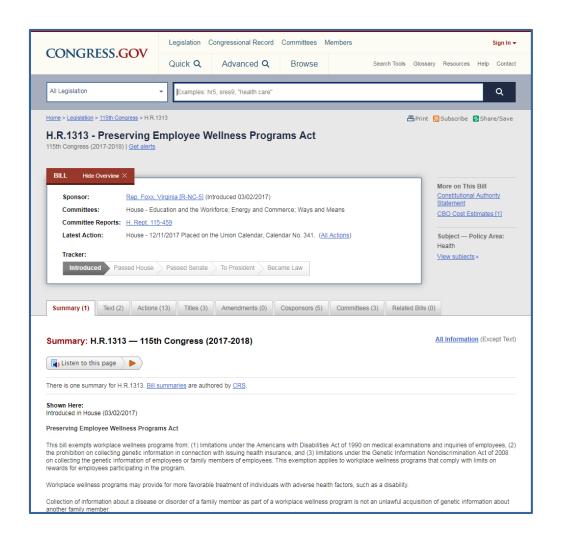
Information protected by GINA

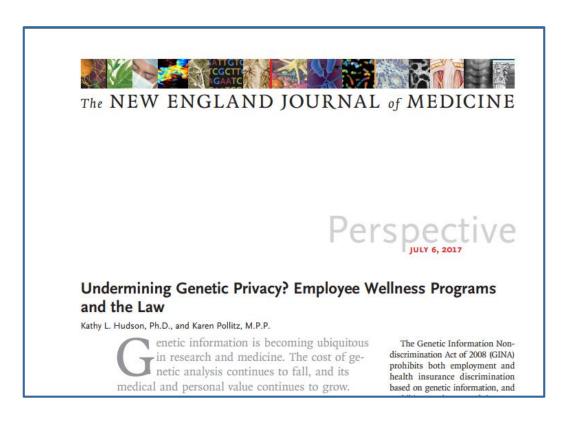
- Family health history
- Results of genetic tests
- Use of genetic counseling and other genetic services
- Participation in genetic research
- □ But....

GINA does not apply to...

- Members of the U.S. military who receive their care through the Tricare military health system
- Veterans who receive their care through the Veteran's Administration
- The Indian Health Service
- Federal employees who get their care through the Federal Employees
 Health Benefits Plans
 - These have their own policies similar to GINA

H.R. 1313 Employee Wellness Program

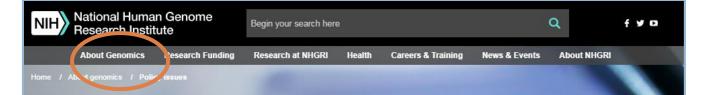




PMID: 28537794

Genomic Health Literacy

- People in a scientifically and genetically literate society are:
 - better able to understand the world they live in
 - better able to understand the diversity of life
 - better able to understand the basics of health and disease
 - more empowered to make informed decisions about their healthcare
 - more empowered to make informed decisions about privacy, security,
 ethical, and societal issues related to genetics



Policy Issues in Genomics

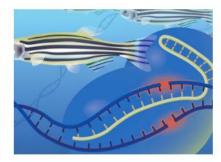
NHGRI is committed to driving the responsible use of genomics in society in order to advance knowledge and ensure that genomics benefits the health of all humans. To do this, we consider the ethical, legal, and social aspect of genomics research in our work, including these key issues.



Coverage and Reimbursement of Genetic Tests >



Genetic Discrimination >



What is Genome Editing? >





Coverage and Reimbursement of Genetic Tests >

Payers such as insurance companies and Medicare need systematic ways of evaluating genetic tests for reimbursement.



Genetic Discrimination >

Begin your search here

In 2008, the Genetic Information Nondiscrimination Act was passed into law, prohibiting discrimination by employers and health insurers.



fyD

What is Genome Editing? >

Genome editing is a method that lets scientists change the DNA of many organisms, including plants, bacteria, and animals.



Health Disparities >

NHGRI wants to ensure that all populations benefit from the advances of genomics research.



Human Subjects Research >

Federally-funded research with human participants must comply with regulations that protect the rights and welfare of the participants.



Informed Consent >

Informed consent shows respect for personal autonomy and is an important ethical requirement in research.



Intellectual Property in Genomics >

In June 2013, the Supreme Court determined that DNA in its natural form cannot be patented.



Privacy in Genomics >

There are laws and policies that serve to protect the privacy of individuals' genomic information.



Regulation of Genetic Tests >

Most genetic tests today are not regulated, meaning that they go to market without any independent

Literacy Resources



discover, educate, advocate,

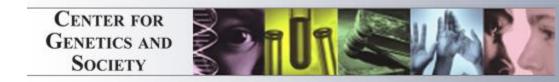


















Precision Medicine

"...a bold new research effort to revolutionize how we improve health and treat disease."

Precision Medicine Initiative

Mission statement:

To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care.

THE PRECISION MEDICINE INITIATIVE



PMI announcement

Precision Medicine is...

- Precision medicine is a revolutionary approach for disease prevention and treatment that takes into account individual differences in lifestyle, environment, and biology.
- Instead of what treatment is right for this disease it is what treatment is right for this patient.

Precision Medicine- Genetics Home Reference



Health Conditions

Genes

Chromosomes & mtDNA

Classroom

Help Me Understand Genetics

▶ Genomic Research

▼ Precision Medicine

What is precision medicine?

What is the difference between precision medicine and personalized medicine? What about pharmacogenomics?

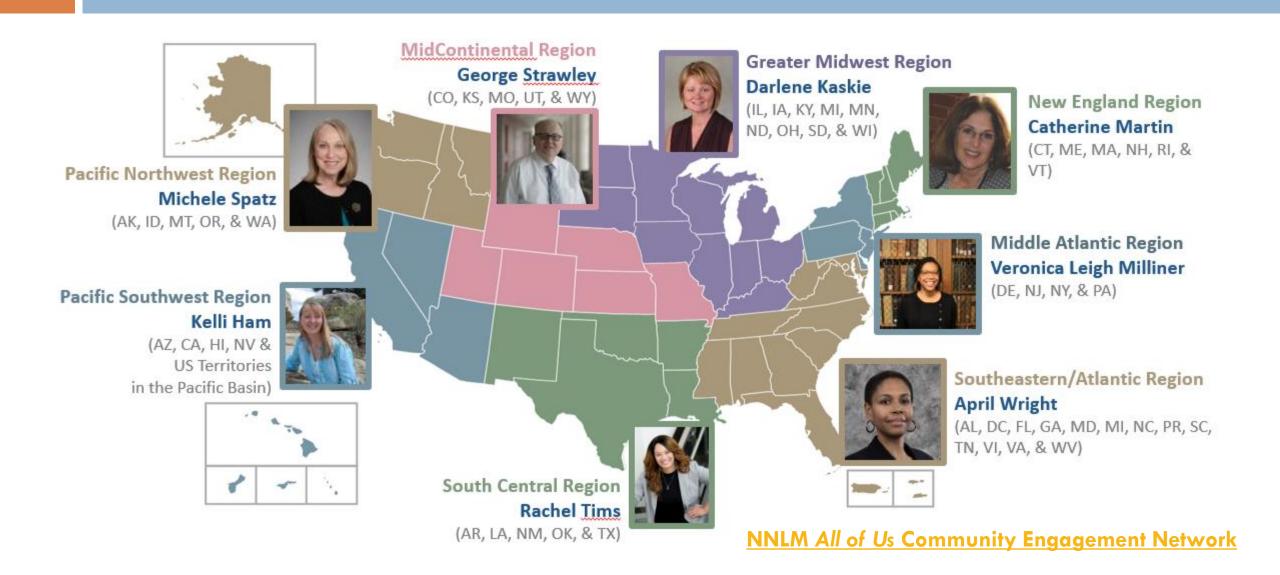
What is the Precision Medicine Initiative?

What are some potential benefits of precision medicine and the Precision Medicine Initiative?

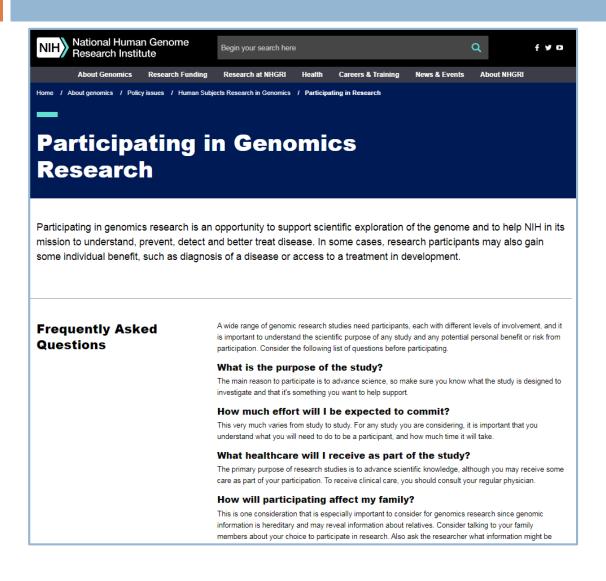
What are some of the challenges facing precision medicine and the Precision Medicine Initiative?

Printable Chapter PDF (1MB)

All of Us & NNLM Community Engagement Team

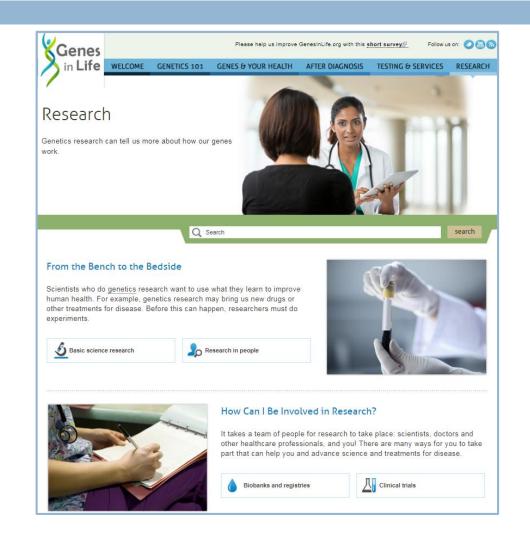


Participating in Genomics Research



- □ What is the purpose of the study?
- How much effort will I be expected to commit?
- What healthcare will I receive as part of the study?
- How will participating affect me and my family?
- Is it physically safe to participate in genomics research?
- How will my privacy be protected?
- Might I be discriminated against as a results of information about my genome being revealed?

Participating in Genomics Research, continued



Participating in research is a choice

Joining a research study is an important personal decision. Before you join, researchers will talk with you about the goals of the study and possible risks and benefits. They will also explain the rules they follow to protect your safety and privacy. Ask for help if you don't understand something or have questions.

You should never feel rushed or pressured to make a decision. Being part of a research study is completely voluntary - it's your choice.

After you understand the study, if you decide to take part you will be asked to sign a document called an "informed consent form." You can change your mind at any time, for any reason, even after you sign.

Ouestions to ask

You have a right to ask questions about genetic research before you decide whether to take part in a research study. You may want to ask the following questions:

- > What is the purpose of the study?
- > How will you collect my genetic sample(s)?
- > Will I receive results from this study?
- > What happens if the tests reveal a medical issue?
- > Will the test results become part of my medical record?
- > Will the genetic testing impact my family?
- > Will test results impact my health insurance coverage in
- > Will my samples or genetic information be used for future research? If so, will I need to give my consent?
- > Will my samples or genetic information be shared with any other researchers?

GENETIC RESEARCH



Genes are part of every cell; they are made of DNA and come in pairs. Genetic researchers often use blood or saliva samples to look at DNA, and they are interested in how genes affect the way our bodies work.









This brochure contains general information for educational purposes and is not intended to provide medical advice. Talk with your own doctor or researc

Questions? Call us at

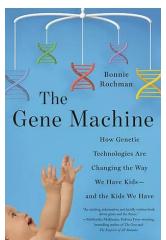
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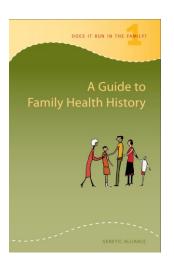
"Preparing the public to make educated personal and family health decisions in a time of rapidly evolving genetic and genomic knowledge will require new partnerships between the education system, health care systems, the government, community advocacy organizations, consumers and the media."

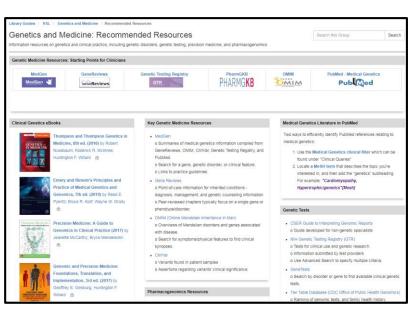
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#thinkbeforeyouspit
#geneticsandhealth
#familyhealthhistory









Show What You Know!

- What is the revolutionary healthcare approach that takes into account individual differences in lifestyle, environment, and biology?
- 2) Clinicians are not concerned about <u>all</u> genetic variants only those that are _____
- 3) Do you need to have a genetic test in order to be prescribed some FDA-approved drugs?
- If you have a known pathogenic variant of BRCA1, will you get breast cancer before you're 80 years old?
- 5) True or False? GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.
- 6) What resource would you recommend to patients who wanted to learn more about a genetic condition?
- 7) What is a good starting place for finding genetic information for clinicians?

Questions?

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